

ABSTRACT BOOK

ORAL PRESENTATIONS

May 30, Thursday

RARE DISORDERS

Session: IMMUNE-MEDIATED DISORDERS

APPROACH TO INITIAL DEMYELINATING EVENT IN CHILDREN

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An inflammatory disorder of the central nervous system (CNS) may underlie any acute or subacute neurological symptoms in a child such as behavioral changes, fever, sleepiness, or focal neurological signs. The first evaluation should include physical examination and testing for inflammatory markers in peripheral blood: many inflammatory CNS events in childhood are part of a systemic disorder, such as vasculitic disorders or hemophagocytic syndromes. On the other hand, the diagnosis of a primary (isolated CNS) inflammatory and/or demyelinating condition is likely in the absence of any systemic organ involvement or serum acute phase reactants. In this case, acute disseminated encephalomyelitis (ADEM), optic neuritis, transverse myelitis, multiple sclerosis (MS), NMO spectrum disorders and, a currently controversial term, clinically isolated episode (CIS), take place in the differential diagnosis. ADEM is more frequent than MS in children younger than 10 years of age. There are no serum biomarkers pointing to ADEM or MS, although antibodies against CNS antigens including myelin oligodendrocyte glycoprotein (MOG) are being investigated. Cerebrospinal fluid oligoclonal bands and IgG index are usually of little help in childhood because of the low rate of positivity in pediatric multiple sclerosis. MRI showing bilateral large lesions with fuzzy borders, subcortical or bilateral thalamic distribution suggest ADEM. Probably the most useful early marker is a follow-up MRI with at least 3 month interval after the initial episode. Treatment in the acute period usually consists of steroids in most demyelinating disorders. An accurate diagnosis is more important for long-term treatment decisions, which have to be taken together with the family and the patient.

MULTIPLE SCLEROSIS IN CHILDHOOD: WHAT ARE THE NEWS?

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Approximately 3–5% of multiple sclerosis (MS) patients experience their first MS attack during childhood. Although pediatric MS closely resembles adult-onset relapsing-remitting MS, it represents several special features: children experience 2–3 times more frequent relapses early in the disease course, approximately one third of children demonstrate evidence of significant cognitive deficits early in the disease course with the worsening on follow up testing at 2 years. Comparative MRI studies have shown a higher T2 lesion burden compared with adults supporting the concept that MS in young age is more inflammatory. Although no pivotal studies of disease-modifying therapies has been conducted, beta-interferons

and glatiramer acetate are widely used in pediatric MS population on the basis of class I evidence in adult MS and a number of class III and IV studies on safety profiles and efficacy outcomes in pediatric MS patients. The International Pediatric MS Study Group (IPMSSG) recommends that all pediatric patients with MS should be considered for treatment with first-line immunomodulatory therapy. Although beta-interferons and glatiramer acetate are generally accepted as the standard of care, in Lithuania it is ongoing debate with authorities about standardized recommendations of treatment of pediatric cases. Despite of early initiation of early treatment the high frequency of inadequate treatment response has been shown in retrospective studies supporting the need to explore the other therapeutic options. In 2012 IPMSSG published consensus on new and existing therapeutics and supported the view that appropriate emerging therapies should be evaluated in pediatric MS with the hope that new efficacious and reasonably safe treatments will be identified.

ACUTE NON-TRAUMATIC MYELOPATHIES

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The term acute myelopathies -referred to a spinal cord dysfunction- represent a heterogeneous group of disorders with distinct etiologies, clinical and radiologic features, and prognoses. Traumatic myelopathies will be no included in this presentation.

Acute myelopathy can be due to several causes: a) infective agents or inflammatory processes, such as in acute myelitis, b) compressive lesions, c) vascular lesions d) toxic agents, e) electrical injury.

The clinical presentation is often dramatic with tetraparesis or paraparesis, sensory disturbances and bladder and/or bowel dysfunction. History and physical examination are used to localize the lesion to the root or specific level of the cord, which can guide imaging. Different syndromes are recognized: complete transverse lesion, central grey matter syndrome, anterior horn syndrome, anterior spinal artery syndrome, etc).

The first priority is to rule out a compressive lesion. If a myelopathy is suspected, a gadolinium-enhanced MRI of the spinal cord should be obtained as soon as possible. If there is no structural lesion such as epidural blood or a spinal mass, then the presence or absence of spinal cord inflammation should be documented with a lumbar puncture.

The absence of pleocytosis would lead to consideration of non inflammatory causes of myelopathy such as arteriovenous malformations, fibrocartilaginous embolism, or possibly early inflammatory myelopathy. In the presence of an inflammatory process (defined by gadolinium enhancement, CSF pleocytosis, or elevated CSF immunoglobulin index), one should determine whether there is an inflammatory or an infectious cause. Different virus, bacterias, parasites and fungi have to be considered as autoimmune and inflammatory diseases that involve the CNS. Illustrative case studies will be discussed.

OTHER CNS AUTOIMMUNE DISORDERS

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The most common disorders causing demyelination in childhood include multiple sclerosis (MS) and acute disseminated encephalopathy (ADEM). However, various other immune-mediated disorders of the CNS other than multiple sclerosis

(MS) and acute disseminated encephalomyelitis (ADEM) exist. These disorders can be broadly grouped into: 1) demyelinating disorders such as Neuromyelitis optica, Schilder Disease and Balo's concentric demyelination; 2) vasculitic and rheumatological disorders that can cause CNS involvement such as Sydenham chorea, systemic lupus erythematosus (SLE), Hemolytic-uremic syndrome (HUS) and Polyarteritis nodosa (PAN), antibody-mediated encephalitis such as NMDA receptor encephalitis.

This lecture will discuss and review the various disorders and provide information in the presentation, diagnosis, and therapies of these disorders.

Session: MUSCLE AND NERVE DISORDERS

OPTIMAL MANAGEMENT OF NEUROMUSCULAR DISORDERS IN RESOURCE POOR SETTINGS

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In most resource poor settings (RPS) the incidence of neuromuscular disease (NMD) is based on estimations. This relates to limited clinical skills to diagnose affected patients, many are mislabelled with cerebral palsy, and lack of access to diagnostic tools such as histopathology and molecular genetics. In reality there are few 'cures' for the hereditary neuromuscular diseases. In developed (resource equipped) settings hereditary neuromuscular diseases dominate but RPS the high incidence of acquired insults (infections, trauma, malnutrition, toxins etc) supports that this group will be more frequently seen. The implication from this is that many of these acquired causes are reversible.

The approach for the management of children with NMD in the author's setting is as follows: A dedicated clinic exists with an attendant physiotherapist. Clinicians are aware of the service and as such awareness has been raised of NMD. Any child thought to be affected or just 'low toned' can be referred to the service. The clinic coordinated the input from multiple other support services including orthotics, orthopaedics, speech therapy, pulmonology and so on. When possible a respiratory technologist and a counsellor, funded by Muscular Dystrophy Foundation, are also part of the clinic. The approach to care is very standard for most types of NMD. Diagnosis is confirmed wherever possible (within the range of neurophysiology, histopathology and molecular genetic services which are available) and any reversible causes targeted in the assessment between acquired and hereditary disease. Once a diagnostic label is made (where possible), and counselling is completed, the optimal chronic care for the child typically consists of maintaining motor function (avoidance of contractures), monitoring respiratory and cardiac function, healthy diet and appropriate orthotic / seating support. Home programs are supported wherever possible. Children living within the region of the hospital are often seen every 3 months. Those further afield have management plans developed and a contact physician guided to follow these. These children are seen at more spaced out intervals. Protocols are developed for the common hereditary NMDs seen in our context – specifically Duchenne Muscular Dystrophy, Spinal Muscular Atrophy and Congenital Myopathies. Most of the peripheral neuropathies patients are recovering severe cases of AIDP (Guillain-Barré syndrome).

For children with NMD, diagnostic closure is important for the genetic and prognostic implications alone. Currently effective 'cures' are lacking, however research is advancing rapidly.

Our policy is that we are attempting get our 'genetic houses' so that if therapies for 'exon skipping' or 'premature stop codons' are found to be effective, we will have data to detect early which patients carry the receptive mutations. For large groups of patients with similar undefined conditions we have successfully collaborated with international centres who have performed complex genetic screens on these patients at no cost to us.

AN UPDATE ON CHILDHOOD HEREDITARY NEUROPATHIES

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Charcot-Marie-Tooth (CMT) disease type 1A is the commonest cause of hereditary peripheral neuropathy worldwide with an incidence of approximately one in 2500. Since the demonstration that CMT type 1A was caused by a duplication of DNA on chromosome 17 which caused an over-expression of the peripheral myelin protein (PMP22) gene, over 30 genes and approximately 20 other loci have been reported which are implicated in causation.

In this presentation, recent advances in the genetics of CMT will be illustrated and an age-related clinical approach will be offered to assist in the correct diagnosis and genetic investigation. Recently-described, potentially treatable variants will be discussed.

THE CHALLENGE OF GUILLAIN-BARRÉ SYNDROME IN CHILDREN

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Guillain-Barré syndrome (GBS) consists in acute peripheral nerve palsy with areflexia and albumino-cytologic dissociation in cerebrospinal fluid (CSF). Although this syndrome is well known, diagnosis may be challenging in a child who stops walking...

GBS is relatively rare in children with a yearly incidence <1/100'000. Nevertheless it is the most frequent cause of acute flaccid peripheral palsy in children. Condition is immune mediated and is preceded by viral or bacterial infections, or vaccination. Lesion concerns the peripheral nerve fiber on its entire length, causing a polyradiculoneuropathy. It provokes motor, sensory and autonomic deficits in proportion that depends on the nerve fibers implicated. Course consists in 3 phases during which symptoms increase, plateau, and then regress. Weakness usually concerns lower limbs initially. It then progresses to the upper limbs, and in some subjects, to bulbar innervation and to the face. Breathing may become impaired and require mechanical ventilation. Recovery varies depending on the proportion and severity of myelinic versus axonal lesion. In GBS that follows infection to *Campylobacter jejuni*, motor axonal lesion dominates, and recovery is delayed.

Rapid disappearance of myotatic reflexes that contrasts with persistence or increase of muscle contraction to direct muscle percussion is an early bedside sign. Sensory symptoms exist in more than half of patients. Increased CSF protein content may be delayed. Nerve conduction studies are helpful in disclosing myelinic lesions (early "indirect double discharges", followed by inhomogeneous conduction slowings and blockings) and also in assessing the degree of axonal loss, thus the prognosis. However, young children do not eas-

ily tolerate the procedure. It may thus be performed under sedation at time of the lumbar puncture.

The challenge of GBS in children is mainly that of the diagnosis since the condition is usually milder and outcome more favorable than in adults.

3-D GAIT ANALYSIS IN CHILD NEUROLOGY

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Background. Accurate 3D gait analysis has been shown in numerous previous studies to alter decision-making in the treatment of children with cerebral palsy (CP), with some authors reporting changes in surgical plans in 89% of patients. Other authors have reported that pre-operative gait analysis decreased the reoperation rate in ambulatory children with cerebral palsy from 40% to 20% by 5 years post-op.

Aim of the presentation. To describe the role and efficacy of gait analysis in the management of ambulatory children with cerebral palsy and to present its use in Kaunas Children's rehabilitation hospital.

Summary. The large number of papers describes studies that analyze the role and the efficacy of 3D gait analysis. According to Hierarchical Model of Efficacy (Wren T., 2011), efficacy of gait analysis can be divided into several levels: technical efficacy (Level 1 – accuracy and reliability of data), diagnostic accuracy efficacy (Level 2 – consistency of interpretation), diagnostic thinking and treatment efficacy (Levels 3–4 – change or reinforcement of decision making), patient outcome efficacy (Level 5 – improved outcomes), societal efficacy (Level 6 – cost effectiveness). Studies show a strong evidence of the effect on decision making in selecting treatment methods in cerebral palsy. They also give evidence that gait analysis improves outcomes of surgical treatment of CP. There are some proves, that gait analysis does not increase costs of services for children with CP, but there are not enough proves that it reduces costs.

Kaunas Children's rehabilitation hospital installed 3D gait analysis equipment a year ago. It took some time to create and train a team that was going to work in gait analysis laboratory. The main problem is, that Sickness Funds refuses to refund expenses related to performing gait analysis. Main patient groups: children with CP and myelomeningocele, traumatic brain and spinal cord injuries. Additional patient groups: elderly and athletes. Despite of financial limitations we believe that 3-D gait analysis will improve habilitation for children with CP and other movement disorders in Lithuania.

Session: METABOLIC/GENETIC DISORDERS

VITAMIN AND COFACTOR RESPONSIVE ENCEPHALOPATHIES AND SEIZURES

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Early diagnosis and treatment of vitamin and/or cofactor responsive encephalopathies and seizures is critical for both seizure control and cerebral development and to prevent the kindling of intractable seizures with secondary brain injury. Recognition of these specific disorders is key to their management given their essential requirement for specific cofactors and their reduced responsiveness to standard anticonvulsant therapy. The overall goals of this talk are; (1) To

provide recognition of the clinical phenotypes of selected treatable metabolic etiologies of early-onset encephalopathies with seizures; (2) To highlight the appropriate diagnostic investigations including the specific biochemical markers, pathology and genetics for each, and (3) To outline the effective treatment strategies including the cofactors/vitamins and specific dietary interventions. The conditions to be reviewed will include the glucose transporter defect (GLUT1); the creatine deficiency disorders namely the X-linked creatine transporter defect, l-arginine:glycine amidinotransferase (AGAT) deficiency and guanidinoacetate methyltransferase (GAMT) deficiency; the serine deficiency disorders namely 3-phosphoglycerate dehydrogenase (3-PHGDH) deficiency and 3-phosphoserine phosphatase deficiency; the biotin-responsive disorders including biotin deficiency and biotinidase deficiency; the folate-responsive disorders including folate deficiency, the folic acid transport defect (SLC46A1), the cerebral folate transport defect FOLR1, and 5,10-methylenetetrahydrofolate reductase (MTHFR) deficiency; and the pyridoxine (Vitamin B6) dependent (antiquitin), folinic acid responsive seizures (FARS) and pyridoxal-5'-phosphate (PNPO) responsive disorders. This will be followed by a sequential approach to unexplained frequent or intractable neonatal seizures.

APPROACH TO A CHILD WITH DEVELOPMENTAL DELAY: A METABOLIC DISEASE?

Linda De Meirleir

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Metabolic diseases can present in a number of different ways. Some are diagnosed after neonatal screening, other present with an acute encephalopathy associated with biochemical disturbances such as acidosis, hypoglycaemia or hyperammonia. Other metabolic diseases will have with a more chronic presentation, including developmental delay, epilepsy and regression.

Associated neurological signs and symptoms such as extra neurological manifestations, psychiatric signs, autistic traits, cerebellar dysfunction and dysmorphic traits can lead to a clue for further specific investigation. Before the age of 5 year we can speak of developmental delay, after that it is defined as mental retardation (MR) or cognitive impairment. Assessment requires a multidisciplinary approach with extensive family history, general physical and neurological examination, including vision and hearing. With the use of the microarray and in some cases whole exome sequencing, genetic tests sometimes precede the metabolic work-up.

In absence of dysmorphism basic blood tests can be recommended including T3, creatine metabolites and urinary GAGS, purines and pyrimidines. The X-linked creatine transporter defect for example can lead to mild to severe MR, associated with expressive speech delay and autistic-like behaviour. MCT8 is another X-linked disorder associating severe mental retardation, with delay in myelination, where high T3 is the marker of this disease. Urinary organic acid analysis can lead to specific diagnosis such as 4-OHbutyric aciduria or L-2-OHglutaric aciduria. In the group of the mucopolysaccharidoses especially Sanfillipo A and B can present with variable behavioural disturbances and cognitive decline without clear somatic changes. The same is true for alpha and beta mannosidosis, being picked up by urinary oligosaccharides analysis. The expanding group of congenital disorders of CDG are often associated with mental retardation, some also with specific MRI abnormalities such as cerebellar atrophy. Many other metabolic disorders will present with cogni-

tive impairment or behavioural changes and might lead to specific diagnosis such as neuronal ceroid lipofuscinosis, Niemann Pick C, X-adrenoleukodystrophy, or cerebrotendinous xanthomatosis and early diagnosis lead to a better outcome with the appropriate treatment.

RECENT ADVANCES IN GENETIC DIAGNOSTICS OF NEURO-PEDIATRIC DISEASES

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New genome-wide approaches have recently changed the field of genetics of human disease. One of the most difficult challenges for clinicians is that the etiology of the disease frequently remains elusive despite costly etiologic investigations. Diagnostic workup is frequently complex due to the large number of genes involved in the etiology of many genetic conditions. The main technologies used for the genetic evaluation of individuals with neurogenetic conditions include chromosomal microarray, specific gene sequencing and next generation sequencing. Currently used whole-genome chromosomal microarrays allow detection of copy number changes of 30–200 kilobases in size. Using this technology, in as many as 20% of patients with intellectual disability, pathogenic microdeletions or microduplications can be identified.

Monogenic neuropediatric diseases can be genetically diagnosed by specific gene and mutation analysis or by parallel sequencing of a large number of genes using a novel technology called 'next generation sequencing'. This technology enables sequencing of all coding genes or even the whole genome in a single test in several weeks for the cost of less than \$2000. Instead of serial molecular testing of one or a few genes, some hospitals introduced a highly multiplexed molecular diagnostic test that enables simultaneous examination of genes associated with more than 500 autosomal-recessive, X-linked and mitochondrial-pediatric diseases. If specific molecular defect in the affected family member is identified, it is possible to offer prenatal or preimplantation diagnosis to at-risk family members.

Unfortunately, only a very small number of genetic diseases can be successfully treated. Therefore, detection of couples at risk of giving birth to a child with a genetic disease is of the utmost importance. At the population level, a large-scale program for the prevention of genetic diseases based on carrier screening can be initiated and successfully implemented in specific populations with a high carrier frequency of specific genetic diseases. The availability of next generation sequencing will enable preventive genetic screening even in populations in which no founder mutations are known.

TUBEROUS SCLEROSIS: WHAT CAN BE DONE TO IMPROVE THE PROGNOSIS?

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Tuberous sclerosis complex (TSC) is a rare autosomal-dominant disorder caused by mutations of two possible genes, TSC1 or TSC2, known as tumour suppressors, responsible for the inhibition of the mTOR (mammalian target of rapamycin) signaling pathways. Mutations in these genes

cause hyperactivation of the mTOR system and result in excessive cell growth and hamartomatous tumours in multiple organs. Skin, brain, kidneys, heart, eyes, lungs may be affected in an age-dependent pattern. The most severe symptoms are early epilepsy complicated by encephalopathy, obstructive hydrocephalus due to subependymal giant cell astrocytoma (SEGA), renal bleeding and hydronephrosis, etc. Recently, novel treatment with mTOR inhibitors proved to be effective in the suppression of progressive proliferative growth of SEGA and renal angiomyolipomas. Clinical trials are ongoing to explore the benefits of this treatment to control other severe symptoms of TSC, like epilepsy, skin lesions, etc. Long-term follow-up of numerous patients is expected to provide more information on the best terms of treatment initiation, duration and other practical issues. Education of medical professionals and families as well as the implementation of management guidelines and the multidisciplinary follow-up of patients at a specialized center is very important to ensure the early diagnosis and prevention of dangerous complications in patients with this rare disorder.

OLD AND NEW CYTOGENETICS IN THE EVALUATION OF INTELLECTUAL DISABILITIES/ DEVELOPMENTAL DELAYS

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According to multiple guidelines, all children with intellectual disability or global developmental delay should have a comprehensive genetic evaluation to elucidate possible etiologies. Specific genetic diagnosis can lead to specific treatment and management, determination of prognosis and recurrence risk and alleviation of psychosocial problems. Chromosome imbalances are the leading cause of intellectual and developmental disabilities. Diagnostics of these abnormalities include several lines of investigations from conventional karyotyping to FISH and to the most advanced genomic microarrays. The diagnostic yield of all these tests can reach up to 50% (Moeschler JB, 2008), especially in cases of associated physical disabilities/dysmorphism.

We present 4 patients from 9 months to 14 years of age sent to the Centre for Medical Genetics, Vilnius University Hospital Santariskiu Clinics with psychomotor retardation/ intellectual disability with or without associated physical disabilities. By applying different cytogenetic and molecular cytogenetic methods all of them were diagnosed with chromosomal abnormalities. Mosaic karyotype with a small additional marker chromosome of unknown origin was identified in a 3.5 years old male patient with a global developmental delay. FISH method was applied for the investigation of a 3 years and 8 months old male patient with a global developmental delay and typical signs and symptoms pointing to Williams - Beuren syndrome (microcephaly, typical facial gestalt, behavioural characteristics, supravalvular aortic stenosis), the diagnosis of the aforementioned syndrome was confirmed. Clinical features of the third female patient at the age of 9 months consisted of developmental delay, corpus callosum agenesis, ventricular septal defect, microcephaly, growth failure, mus-

cular hypotonia, mild dysmorphic features, diastasis lineae albae, right talipes equinovarus. A large terminal 1q42.3-1q44 deletion involving more than 13 Mb was diagnosed by applying FISH for subtelomeric chromosomal regions, later it was confirmed by comparative genomic hybridization method. The fourth patient, 14.5-year-old female, had a mild intellectual disability, internal hydrocephalus, gait and coordination abnormalities, scoliosis, contractures of ankle joints, and facial dysmorphism. A 7p22.1 de novo microduplication, 1 Mb in size, was detected with the help of array comparative genome hybridization, later characterized as a novel microduplication syndrome (Preiksaitiene E. et al, 2012).

Conclusion: extensive application of various cytogenetic and molecular cytogenetic methods for the genetic evaluation of patients with developmental delay/ intellectual disability provides an opportunity to establish specific diagnosis and to characterize novel genetic syndromes due to chromosomal imbalances.

May 31, Friday

Session: DEVELOPMENTAL ISSUES AND STROKE

HUMAN BRAIN DEVELOPMENT AND PLASTICITY

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We previously demonstrated, using positron emission tomography (PET) and 2-deoxy-2(18F)fluoro-D-glucose (FDG), that neonatal brain glucose metabolism is highest in primary sensory and motor cortex, cingulate cortex, medial temporal region, thalamus, brainstem and cerebellar vermis. Increases of glucose utilization are seen by 2–3 months in parietal, temporal and primary visual cortex, basal ganglia, and cerebellar hemispheres. Between 6 and 8 months, lateral and inferior portions of frontal cortex become more functionally active and, between 8 and 12 months, the dorsal and medial frontal regions also show increased activity. These changes in brain glucose metabolism correlate well with behavioral maturation of the infant. By approximately one year, the infant's pattern of glucose utilization resembles qualitatively that of the adult. However, cortical metabolic rates of glucose utilization follow a nonlinear profile which is consistent with periods of synaptic exuberance and pruning in the human. These patterns of brain maturation provide some perspectives on developmental plasticity periods and timing of interventions, as well as the onset of some pathological processes.

In children with early unilateral injury to the occipital cortex (e. g., in Sturge-Weber syndrome), the contralateral (normal) occipital cortex shows increased glucose metabolism presumably due to reorganization of the optic radiation. Indeed, this has been shown using diffusion tensor tractography. In children who have undergone cerebral hemispherectomy due to intractable epilepsy, both PET and diffusion tensor MRI show reorganizational changes in gray and white matter.

The early development of limbic brain structures as shown on PET scans led us to study children who had suffered from early socioemotional deprivation. On FDG PET scans of post-institutionalized Romanian orphans, we reported decreases of brain glucose metabolism in limbic structures, in-

cluding amygdala, hippocampus, orbital frontal and infra- limbic prefrontal cortex, lateral temporal cortex, and brainstem. These brain regions form networks that are activated by stress and damaged with prolonged stress. More recent studies using diffusion tensor MRI have found abnormal connectivity in uncinate fasciculus, which projects between medial temporal lobe structures and orbital frontal cortex. Such neuroimaging studies provide an unprecedented opportunity to further understand the biological basis of early social deprivation and attachment disorders, and may be also useful to monitor therapeutic interventions.

THE BURDEN OF NEURODISABILITY IN THE WORLD

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Recent estimates of the global burden of disease have highlighted the lack of data on neurodisability, particularly in resource poor countries. The data that is available suggest that the burden is greater than in resource rich countries. With the improved survival in many regions, this burden is likely to increase.

The World Health Organization (WHO) definition of impairment includes a molecular, cellular, physiological or structural disorder, whilst disability is the stable and persistent deficit in function, often the consequence of impairment. The WHO estimates the global burden of diseases with the Disability Adjusted Life years (DALYS). DALYS is the sum of Years Lost of Life (YLL) plus the Years Lost to the Disability (YLD), where YLD is the product of incidence, duration and disability weight. Thus a condition that has a high incidence with severe disability, in which the affected individuals survive for a many years results in a considerable burden.

Estimation of the burden of neurodisability in resource poor countries is hampered by lack of reliable medical records, poorly resourced health facilities and absence of equipment to assess some forms of impairment eg hearing, and the lack of culturally appropriate tools to assess cognitive impairment. Most of the data on impairment comes from relatively small surveys of neurodisability or neuroimpairment conducted on children or specific conditions eg stroke in adults.

Systematic reviews have identified large gaps in our knowledge, in particular behavioural outcomes. Most children have multiple impairments, and the impact of these impairments combined are difficult to assess.

Despite the lack of data on neurodisability in resource poor countries, it appears that it is more prevalent than the resource rich countries, and will increase with improved survival. Further research on the epidemiology and causes of neurodisability is required, to guide measures to prevent disability in these regions.

NEW PERSPECTIVES OF CEREBRAL PALSY FROM A POPULATION-BASED REGISTRY

Michael Shevell

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A population-based registry provides the best opportunity to minimize ascertainment bias to capture an accurate profile of the phenotype of any condition. Utilizing a population-based registry established over a decade ago in the province of Quebec, Canada detailed information will be presented on the following; epidemiologic profile, neurologic sub-types, gross

motor severity (GMFCS), fine motor severity (MACS), comorbidities spectrum and neuroimaging findings. The inter-relationships between these complementary ways of describing and delineating this disorder will also be presented. By detailing a more accurate profile of this neurodevelopmental disability, important decisions regarding service provisions and clinical care can be made at a policy level on a more rational and evidence-based basis.

CHILDHOOD STROKE AND DIFFERENTIAL DIAGNOSIS

Vijeya Ganesan

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This talk will focus on current clinical concepts relating to the diagnosis and management of acute stroke, in particular arterial ischaemic stroke, in children.

Vascular stroke syndromes are as common as brain tumour in children, in the top 10 causes of childhood death, with resultant disability in 2/3rds of survivors. Acute stroke is defined by the WHO as “an acute focal neurological deficit of likely vascular origin”. However, in children, the differential diagnosis of acute focal neurological deficits is wide and at least 1/3rd of children with such presentations will have a non-vascular aetiology. These “stroke mimics” include acute infection, demyelination and space occupying lesions. Thus, imaging is crucial for distinguishing between vascular and non-vascular causes of acute focal deficits, and between arterial ischaemic stroke (AIS; cerebral infarction in an arterial distribution), cerebral venous thrombosis and intracranial haemorrhage. Magnetic resonance imaging enables non-invasive structural and physiological brain imaging, differentiation between AIS and “stroke mimics” and non-invasive evaluation of the cerebral circulation.

Arterial ischaemic stroke in children (>28 days of age) affects children with another medical diagnosis in around 50% of cases. These include sickle cell disease, congenital heart disease, childhood malignancy as well as a number of genetic and syndromic conditions. Sickle cell disease (SCD) is the commonest risk factor world-wide; in people with SCD high middle cerebral artery velocities on transcranial Doppler ultrasound predict those at highest risk of AIS and primary prevention by prophylactic blood transfusion is extremely effective. Of other AIS risk factors, the most important is non-atheromatous cerebral arteriopathy, that is identified in 60–80% of people. The most common morphology is focal occlusive disease of the proximal middle cerebral/distal internal carotid artery – currently termed focal cerebral arteriopathy of childhood. Other entities are dissection (especially cervical), moyamoya (occlusive ICA disease with basal collaterals) and cerebral vasculitis. The use of standardized consensus disease definitions to describe cerebral arteriopathies (e.g. Sebire et al 2004, Bernard et al 2011) is to be encouraged to facilitate sharing data for clinical and research purposes, and current disease definitions are largely based on MRA features. The morphology and course of cerebral arteriopathy is the most important predictor of recurrence and determines secondary prevention. In particular, ominous radiological signatures are the presence of “moyamoya” collaterals, and of progressive arteriopathy.

There are currently no acute treatments for childhood AIS. Thrombolysis (intravenous/intra-arterial) is of unproven benefit and likely to have a role only in a very small minority of children. The mainstay of acute care is to maintain optimal homeostasis (oxygenation, temperature). Exchange transfusion is recommended for AIS in SCD. The mainstays of secondary prevention are anticoagulation or antiplatelet agents; both are

unproven and there is some difference in practice internationally as to choice of agent. In the UK, anticoagulation is reserved for cardioembolic stroke and extracranial arterial dissection. Patients with SCD are offered long-term blood transfusion, those with moyamoya are evaluated for surgical revascularization and those other focal arteriopathies are maintained on aspirin.

PAEDIATRIC STROKE IN ESTONIA: EPIDEMIOLOGY AND RISK FACTORS

Rael Laugesaar, Anneli Kolk, Tiina Talvik

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Stroke is an increasingly recognised cause of childhood mortality and long-term neurological morbidity. The number of studies on paediatric stroke has increased over the past decades due to increased recognition and the possibilities of modern neuroradiology. Many disorders and risk factors have been associated with paediatric stroke but the pathogenesis of stroke in children often remains unclear.

The study was designed to investigate the incidence rate of paediatric stroke in Estonia and to evaluate the risk factors of paediatric stroke. Additionally, we studied the outcome of perinatal stroke, time lag to the diagnosis of childhood arterial ischemic stroke and the association between Factor V Leiden and prothrombin 20210G>A and paediatric ischemic stroke.

Epidemiological studies included 38 children with perinatal stroke and 48 children with childhood stroke. The case-control study included 75 patients and 400 controls. In all cases, the diagnosis of stroke was confirmed by neuroradiological studies (MRI or CT).

Our study revealed that the incidence rate of perinatal stroke in Estonia is 63 per 100,000 live births, which is higher than estimated in previous studies in other countries. The incidence rate of childhood stroke in Estonia is 2.73/100,000, which is similar to earlier reported data. One-third of patients with perinatal stroke have symptoms (most often seizures) after birth (neonatal stroke). The remaining two-thirds reach medical attention at a mean age of eight months, mostly because of hemiparesis (presumed perinatal stroke). Most frequently identified risk factors of perinatal stroke are primiparity, emergent caesarean section, an Apgar score <7 at the first minute, preeclampsia and prothrombotic factors. Arteriopathy, cardiac disorder and prothrombotic factors are the main risk factors of childhood arterial ischemic stroke. Hemiparesis occurs in all children with perinatal stroke at outcome. Epilepsy develops in one-third of children, both with neonatal and presumed perinatal stroke. The diagnosis of childhood arterial ischemic stroke is delayed at average 9.2 days. Children carrying factor V Leiden or prothrombin 20210G>A mutation have 3.1-fold increased risk to have sinovenous thrombosis.

Session: EPILEPSY 1

RE-CLASSIFICATION OF EPILEPSY: A TRUE CHALLENGE?

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In the course of the 20th century a number of successive taxonomic classifications for the epilepsies and for epileptic seizures have been proposed and -temporary- accepted by the ILAE, WHO and the epilepsy community at large. Each of

these tried as much as possible to incorporate the at that time newest scientific findings and concepts.

Due to the fast development of new scientific knowledge and concepts as contrasted to the slow and bureaucratic process of creating a new classification that is acceptable to all and can be practically implemented in general practice, even the latest and formally still official ILAE classifications of the Epileptic Seizures (1981) and Epilepsies and Epileptic Syndromes (1989) were to some extent already outdated by the time of their acceptance. New findings, notably in the field of Genetics, Imaging, Molecular Neurobiology and Extra- and Intracranial EEG & Video Seizure monitoring have shown that the simple and easy applicable double dichotomy: 'focal' vs. 'generalised' and 'idiopathic' vs. 'symptomatic' is an oversimplification, that the addition of 'Cryptogenic' has resulted in a major transatlantic misunderstanding, that 'impairment of consciousness' is not necessarily associated with automatisms and is difficult to assess anyway, etc. Moreover the linguistic problems created by words like 'generalised' that has multiple meanings -anatomical, neurophysiological and semiological- have become increasingly urgent.

New approaches such as 'an semiological classification' or a 'multi-axis diagnostic system' have shown to have some value but have not resulted in a true new classification. A more recent approach has been the redefinition of older terms, notably for 'generalised' versus 'focal' seizures, the etiological concepts regarding the various epileptic syndromes and the introduction of a new concept, 'system epilepsies' that in part seems to apply to the older 'idiopathic' epilepsies but implies a much better defined pathophysiological and neuroanatomical system to be involved.

While the specialists meet and argue the clinicians are waiting; many expressing the need for a practical diagnostic system rather than a classification, especially where this new classification still seems far away and, even when proposed and accepted, will take years to become the standard in clinical practice at large.

Our redefinitions of the seizures and the epilepsies are well on their way and certainly will be helpful for the real specialists in their thinking about and dealing with epilepsy and persons with epilepsy. Classification however remains a next step and for the time being still a true and difficult challenge.

SUPER-REFRACTORY STATUS EPILEPTICUS: TREATMENT WITH KETOGENIC DIET IN PEDIATRICS

Guillermo Agosta

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Introduction. Super-refractory status epilepticus is the type of status which persists despite suitable treatment with multiple anti-convulsive schemes, including prolonged coma with general anesthetic. Different pharmacological treatment schemes have been proposed in these patients, including the use of a Ketogenic diet.

Patients and methods. We studied five records of children from 1 to 14 years of age, in a retrospective analysis, three of whom were diagnosed with FIRES (Febrile Infection-Related Epilepsy Syndrome) and two with a diagnosis of refractory symptomatic partial epilepsy. The average age was six years and the average duration of the status epilepticus was 32 days.

Results. All the patients were given multiple therapeutic schemes, in which pharmacological coma with barbiturates was obtained to reach paroxysm-suppression pattern on electroencephalogram. Since the results of these strategies

were not successful, a classical Ketogenic diet was indicated. After starting the Ketogenic diet, the clinical and electroencephalographic status epilepticus ceased in four patients with good tolerance. One patient did not respond and died.

Conclusions. In patients with super-refractory status epilepticus, when different anticonvulsive schemes are unsuccessful, the Ketogenic diet would be a good option. The Ketogenic diet in this severe clinical situation is highly effective and safe.

RISK FACTORS AND EARLY MULTIDISCIPLINARY MANAGEMENT OF CHILDREN WITH NON-EPILEPTIC SEIZURES

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Objectives. To present new data from the first multisite sibling controlled study on child and family related risk factors in pediatric NES and the clinical implications for the NES assessment and treatment in children. To discuss how early multidisciplinary management can provide foundation for successful transition to psychiatric treatment.

Background. Pediatric NES presents a complex clinical dilemma because of the difficulties in timely diagnosis and lack of evidence based treatment resulting in significant variability in NES management. As a result, youth with NES are often undiagnosed and untreated, and subject to increasing medical and psychiatric morbidity. High rates of psychopathology, specifically depressive and anxiety disorders, learning, attention problems, and academic underachievement were reported in retrospective studies of children with NES, despite of their average IQ scores. Children with NES report their most common stressors as conflicts with teachers, problems at school and at home. Poor coping with stress has been associated with somatic symptoms in children, including high anxiety sensitivity, somatic preoccupation, and functional disability in children with NES.

Although no standard NES treatment practices are available, it has been suggested that early multidisciplinary intervention, started at the epilepsy monitoring unit after the initial NES diagnosis, provides NES youth and their families with necessary knowledge about NES and its management, support, and clinical services needed for successful transition from neurology to psychiatric treatment.

Results. The prevalence, severity, and specificity (conversion, anxiety, and depression, learning disorders) of psychopathology in NES children and its relationship with epilepsy will be discussed. Also, high prevalence of psychiatric diagnoses in the siblings of NES children will be discussed, which endorses the importance of familial psychopathology in the development of NES. Contrary to our hypothesis, there was no association between epilepsy and psychopathology in the NES group. This finding emphasizes that children with NES, with or without epilepsy, have severe mental health problems.

Specific early management strategies, used at Lurie Children's Hospital of Chicago will be presented and discussed.

Conclusions. The methods, findings, and discussion of the study presented in this symposium will highlight the importance of early identification of comorbid psychopathology and risk factors, as well as evidence-based treatment of pediatric NES that should start with early multidisciplinary intervention.

NEONATAL SEIZURES

Ahmed Raouf

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Neonatal seizures are abnormal electrical discharges in the CNS of neonates usually manifesting as stereotyped muscular activity or autonomic changes.

The immature brain seems more prone to seizures than the more mature brain. Seizures are more common in the neonatal period than during any other time throughout life. The incidence of seizures in infants born at term is 1.5–3.0 per 1000 live births; the incidence is even higher in preterm infants, ranging from 50–150 per 1000 live births (Ronit M Pressler, 2005. NSE).

Neonatal seizures often are a manifestation of significant neurologic disease and a major predictor of adverse neurologic outcome in the newborn. The clinical features and electroencephalographic (EEG) characteristics of neonatal seizures differ considerably from those associated with epilepsy in older infants and children, an observation that reflects the immature stage of development of the newborn brain. Another major difference relates to the fact that neonatal seizures rarely are idiopathic. Prompt diagnosis, investigation to establish the underlying etiology, and rapid intervention are essential to minimize the possibility of associated cardiorespiratory instability and to correct treatable causes. Furthermore, experimental data suggest that ongoing or prolonged seizures may cause additional cerebral injury and have detrimental long-term effects.

This presentation is going to show video cases with differentiation of most common neonatal epileptic disorders with non epileptic cases.

MELATONIN SECRETION-EXCRETION PATTERNS IN CHILDREN WITH EPILEPSY

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Melatonin (MLT) is a pineal gland hormone that is synthesized from tryptophan and released in circadian rhythm, with peak concentrations at night. Its role in epilepsy has been noted to the epileptogenic process directly (g-aminobutyric acid (GABA)) neurotransmission, oxidative stress, or to the exacerbations of seizures via disrupted sleep patterns.

The main objectives of the present study were: i) to determine the most frequent kinds of sleep disorders in children with epilepsy; ii) to characterize the melatonin (MLT) system in children with epilepsy in detail: to describe full diurnal profile, to search correlation with anthropometric data, seizure characteristics (time, type of seizures, antiepileptic medications), as well as the other physiological circadian rhythms (body temperature, pulse, blood pressure).

The study results have shown that sleep disorders were more frequent in the group of children with epilepsy (Sleep disorders scale for children (SDSC) (Bruni O et al, 1996)). Melatonin metabolism (AUC/per kg body weight) had negative correlations with age and sexual maturity stage (Tanner stage) in each group separately and in pooled total group. The diurnal profiles of MLT secretion and excretion showed high inter-individual variability in both study groups. The circa-

dian rhythms of salivary MLT secretion and excretion of metabolite 6-sulphatoxymelatonin (aMT6s), core body temperature, pulse, blood pressure were preserved in children with epilepsy. In the group of children with epilepsy, a subgroup of children had very high peak nocturnal salivary MLT concentrations (9/50; 18%). MLT concentrations showed no associations with seizure characteristics. The study suggests that MLT system should be studied further in children with epilepsy and evaluated when treatment with melatonin is intended.

Session: EPILEPSY II

COGNITIVE OUTCOME OF INFANTILE SPASMS: DOES THE TREATMENT HAVE AN EFFECT?

Raili Riikonen

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The following topics are here reviewed: 1. Does the successful short-term treatment of infantile spasms lead to long-term improvement of cognitive outcome? 2. The role of vigabatrin (VGB) in 'other symptomatic group' (excluding tuberous sclerosis). 3. The role of topiramate, valproate, zonisamide, levetiracetam, sulthiame in the treatment. 4. Role of different steroids. 5. Role of early intervention.

Many studies have shown that short duration of hypsarrhythmia (short delay for treatment and early response) is important for cognitive outcome. Steroids will bring more patients and faster to a response, and to better cognitive outcome, at least in patients with cryptogenic spasms.

VGB is a drug of 1st choice in tuberous sclerosis. However, in the 'other symptomatic group', it has only a modest effect of 23% (6 studies), in contrast to ACTH 65% (2 studies).

The evidence is insufficient to recommend the use of topiramate, valproate, zonisamide, levetiracetam, sulthiame, or ketogenic diet for treatment of infantile spasms.

The evidence is insufficient to recommend prednisolone, dexamethsone, and methylprednisone for treatment of infantile spasms. However, high doses of prednisolone has recently shown to bring a high response rate (64%, 4 studies).

The health carers do not recognize infantile spasms. The favourable outcome is associated with short treatment lag.

Conclusions: The successful short-term treatment of infantile spasms may lead to long-term improvement of cognitive outcome.

EPILEPTIC ENCEPHALOPATHIES: FROM CHILDHOOD TO ADULTHOOD

Marina Nikanorova

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Recent years have shown an increase of the life expectancy and social integration of patients with epileptic encephalopathies. Although the treatment goals remain the same regardless of age, the clinical symptoms, EEG-abnormalities, mental and behavioural aspects, side effect profiles of antiepileptic drugs change over time, requiring evolving and sometimes innovative therapies. This presentation will cover the electro-clinical evolution of epileptic spasms, Dravet syndrome and Lennox-Gastaut syndrome.

Besides classical West syndrome, epileptic spasms can occur in other epilepsies of childhood – severe multifocal epilepsy with independent spike foci, Lennox-Gastaut syndrome,

some genetically determined epileptic encephalopathies (CDKL5, STXBP1 mutations) and others. Moreover, epileptic spasms can first manifest in adulthood.

Adult patients with Dravet syndrome present as handicapped persons, always slow in movements, language and thinking. Epilepsy is severe, with frequent nocturnal GTCS, often fever sensitive, but atypical absences and myoclonic seizures are rare. The EEG usually does not display any specific abnormalities. These patients are mentally retarded, sometimes psychotic, with a constant motor impairment: ataxia, myoclonus, clumsiness. They can have severe orthopedic problems, and many of them live in the specialized institutions.

Lennox-Gastaut syndrome (LGS) is one of the most severe and unfavourable epileptic conditions. The underlying mechanisms of LGS remain to be elucidated and the prognosis is not significantly improved despite the development of new therapeutic options. In the majority of cases LGS has a chronic course, and in approximately 50% of patients, it evolves in adulthood into non-specific generalized or multifocal epilepsy. The tonic seizures persist and are the predominant seizure type with time, whereas atypical absences and myoclonic seizures may subside. Moreover, the diffuse slow spike-waves on the EEG frequently disappear, while rapid rhythms during sleep are constant EEG-finding in adult patients. The mental deficit progresses over time, despite the decrease of seizure frequency and some EEG improvement.

Thus, severe epileptic encephalopathies with childhood onset constitute life-long diseases, and in many cases their electro-clinical features change over time. Most of these conditions are refractory to treatment and are associated with mental deterioration. However, the early treatment with new potential antiepileptic drugs can provide the better seizure control and more favourable cognitive outcome.

CSWS, ESES, LKS – A CONFUSION IN TERMINOLOGY

Orvar Eeg-Olofsson

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'Continuous spikes and waves during sleep' (CSWS) and 'electric status epilepticus during sleep' (ESES) have been used without discern when referring to affiliated but different entities. A North American survey (Fernandez et al, 2012) exposed the heterogeneity in defining these concepts among pediatric neurologists and epileptologists. The results will be the same in Europe. In spite of the fact that 'ESES' should be used for the EEG pattern and 'CSWS' for an epileptic encephalopathy there are frequent misconceptions in the literature. That the EEG pattern should consist in at least 85% of the NREM sleep tracing is wrong. There is now evidence that less epileptiform activity can be found in typical 'CSWS'. The pattern is found in a spectrum of electroclinical syndromes and neurodevelopmental disorders. The distinction in phenotype expression in this spectrum can be explained by maturational and genetic differences, and by the physiologic characteristics of the dysfunctional brain area. The confusion has deepened as the Landau-Kleffner syndrome (LKS) has sometimes been considered equivalent to 'CSWS' with reference to the EEG pattern. However, in 'LKS' mainly posterior temporal and parietooccipital foci of epileptiform activity is found. Sometimes the pattern is multifocal or bilateral synchronous and can show up as 'ESES'. Regarding terminology 'ESES' could be replaced with 'frequent spike-waves during sleep' (FSWS). 'CSWS' could be called 'electroclinical syndrome with various seizure types, cognitive regression and FSWS'. 'LKS' should be equivalent to 'electroclinical syndrome with acquired aphasia'. FSWS must be characterized

with respect to quantification and lateralization. A cut-off value is not of interest as quantification using calculation of a spike index will provide all information. It is important to heighten the discussion toward increased awareness of terminology in order of homogeneity.

IDIOPATHIC PARTIAL EPILEPSY AND LANGUAGE DYSFUNCTION. FROM ROLANDIC EPILEPSY TO ACQUIRED EPILEPTIC APHASIA

Thierry Deonna

Unité de Neurologie et de Neuroréhabilitation Pédiatrique, Univ. Children's Hospital, CHUV, LAUSANNE, Switzerland

This lecture is intended as a journey through the history of ideas, conceptual changes and accumulating data of all sorts of horizons, including genetics, modern electrophysiology and brain imaging, related to the above title. This theme is occupying an always larger number of different professionals, with increasingly specialized and sophisticated background, often far from the clinical field. Early travelers who charted the way to modern childhood epilepsy in the 1970's and those whose main country was the development of language and cognition rarely met on the same roads, except in polite formal encounters as invited guests in professional congresses.

The recognition that there is a close link between the idiopathic-genetic partial epilepsies of childhood (rolandic epilepsy being the most frequent presentation) and those with acquired cognitive manifestations (acquired epileptic aphasia being the historical prototype) was long to come. The hallmark of these idiopathic partial epilepsies is a strong preponderance to involve the perisylvian regions, regions that are naturally predisposed to process the very specialized language components, both in comprehension and production, any of which can be affected by the epileptic process in isolation.

There is also increasing suspicion that minor learning disabilities or even developmental or behavioral problems in these otherwise "benign epilepsies" are not only due to some developmental brain immaturity or structural brain deviation, but can be also "epilepsy-related" (that is due to focal paroxysmal EEG epileptic activity with often major sleep activation). This remains hard to determine in an individual case, although there is much indirect evidence that it does happen. In some of these children, spontaneous "normalization" of specific delayed developmental functions has been documented in longitudinal studies, whereas others persist or even aggravate, particularly during unusually active or severe epilepsy periods. Furthermore, the regression of an initially delayed but previously fully compensated for competence (typically language) can also occur later on for the same reasons. With this complex background in mind, it is a major but necessary challenge to consider the introduction and evaluate the possible positive effects of antiepileptic drugs (and which) on cognitive functions.

SLEEP AND BEHAVIOURAL PROBLEMS IN ROLANDIC EPILEPSY

Ruta Samaitiene

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Background. Although patients with benign childhood epilepsy with centrotemporal spikes (rolandic epilepsy) exhibit a

benign course of the disease, some of them display sleep and behavioural problems.

Methods. Seventy-five patients with rolandic epilepsy, aged 6–11 years, were included in this study. The patients were divided into two subgroups according to the presence of seizures over the preceding 6 months. The comparison group comprised 32 patients without epilepsy and with similar characteristics in terms of age and sex. All patients underwent evaluation of sleep (Sleep Disturbance Scale for Children, SDSC) and behaviour (Lithuanian version of Child Behaviour Checklist, CBCL).

Results. Only patients who had had seizures over the preceding 6 months displayed significantly higher scores for sleep problems (sleep breathing disorders, excessive daytime sleepiness, disorders of sleep-wake transition), worse sleep quality (longer sleep onset latency), and behavioural problems (social problems, thought problems, attention problems, and aggressive behaviour) than the patients of the comparison group. Presence of additional extrarolandic focus and focal specific discharges spreading to centrofrontotemporal, centroparietotemporal areas were related to the higher CBCL scores and treatment with second line AED. Parents' ratings for existing sleep problems were sensitive to CBCL and SDSC scores above normal values.

Conclusions. Children with active rolandic epilepsy are at risk for sleep and behavioural problems. Presence of discharge location other than centrotemporal was related to higher scores of behavioural problems and treatment with second line AED.

June 1, Saturday

Session: MISCELLANEOUS I

ICNAPEDIA: THE CHILD NEUROLOGY KNOWLEDGE ENVIRONMENT

Biju Hameed

University of Bristol, United Kingdom

ICNAPedia is the knowledge environment portal and global networking platform published by the ICNA with the purpose of achieving its stated aim of promoting education and research in child neurology worldwide. Utilizing the latest social networking, database mining tools and content management technologies, ICNAPedia enables child neurologists and allied professionals to connect, collaborate with and benefit from mutual knowledge transfer.

The goal is to realize a more distributed structure of knowledge transfer, particularly suited to the infrastructure in developing countries. ICNAPedia also serves to facilitate research, audit and surveillance in child neurology. ICNAPedia regularly feature highlights from professional focused events around the globe, concise news related to child neurology and byte size information from the vast amount of literature being published daily from around the world. By delivering key content in a clear concise way, it aims to combat information overload.

In today's globalised world, these tools will enable the physician to enhance their professional relationships, gain knowledge and insight into the practice of child neurology around the world and actively contribute to the advancement of health care for children with neurological problems and neurodisability.

ATAXIAS IN CHILDHOOD: DIFFERENTIAL DIAGNOSIS AND MANAGEMENT

Robert Rust (USA)

University of Virginia School of Medicine, Virginia, USA

Ataxic conditions are those that interfere with the localization of body parts in space, most strikingly the arms, legs, trunk and eyes. Abnormal function may be the consequence of abnormal sensory input to the cerebellum or to dysfunction of the cerebellum itself. There is a developmentally determined segregation of the cerebellar functions. Examples of these aspects of ataxia will be discussed. Ataxias may be acute in onset without further development of the ataxic condition as may occur in stroke related ataxia. Other causes of ataxia; infectious, inflammatory, vascular, toxic, heritable, and so forth, ataxias may demonstrate clinically and historically associated features that are helpful in diagnosing and treating the particular type of ataxia. Some ataxic conditions may have a very long clinical evolution as is particularly true in genetically determined ataxias. Abnormalities of the function of the extrapyramidal system or of the brain stem, and in some instances hearing are additional important indicators at the type of ataxia and of its cause. Ataxia may also be psychogenic. Toxins as well as trauma may induce ataxia. Ataxia is not the consequence of cerebellar paroxysms since the functions of the cerebellum are to eliminate excessive function rather than to produce positive functional consequences. There are however treatable causes of ataxia, chiefly inheritable metabolic diseases. This talk will focus on organizing the approach to ataxias since there are too many conditions to consider independently. However, some particularly pertinent causes of ataxia in children will be discussed as examples of this approach.

CHRONIC DAILY HEADACHE IN CHILDREN

Kenneth J. Mack

Mayo Clinic, Rochester, Minnesota, United States of America

Chronic daily headache (CDH) occurs in 1–2% of children and adolescents. It can evolve from either episodic tension-type headache or episodic migraine, or can appear with no previous headache history. As with other primary headache disorders, treatment is based on the level of disability. There are children and adolescents who cope well, but there are others who are markedly disabled by their chronic headaches. As in adults, children and adolescents with CDH are at risk for medication overuse.

CDH is a diagnosis of exclusion, based on a thorough history, normal physical examination, and negative neuroimaging findings. Along with the chronic headaches, children with this condition may have co-morbid sleep problems, autonomic dysfunction, anxiety, and/or depression. Principles of treatment include identifying migrainous components, stopping medication overuse, stressing normalcy, using rational pharmacotherapy, and addressing co-morbid conditions. Successful outcomes often involve identifying an appropriate headache preventative, reintegration into school, and family participation in resetting realistic expectations.

RECENT ADVANCES IN THE MANAGEMENT OF ACUTE BACTERIAL MENINGITIS

Pratibha Singhi

Rehabilitation Centre Chandigarh, India

Acute bacterial meningitis (ABM) is an important cause of mortality and morbidity particularly in developing countries. Early diagnosis and prompt treatment with appropriate antibi-

otics and adjunctive therapy are important. Although clinical symptoms and signs suggest the diagnosis, CSF analysis is required to make a specific diagnosis and for organism identification and isolation. In children who have received prior antibiotic therapy, the yield of Gram stain and culture may be reduced. Rapid diagnostic tests and PCR are particularly helpful in such cases. CT Scan before LP is not routinely required; it is indicated if there are symptoms and signs of raised intracranial pressure or a mass lesion. Broad spectrum empiric antibiotic therapy according to the age of the child and underlying predisposing condition is started before isolation of the pathogenic organism. Definitive antibiotic therapy is tailored to the causative organism and its pattern of resistance. Corticosteroids given before or along with antibiotics reduce hearing loss in *H. influenzae* or Pneumococcal Meningitis but do not increase survival. Their utility in children from resource poor countries is not established. Oral glycerol for first 48 hours has been shown to reduce the risk of neurological sequelae in ABM in some studies. Normal maintenance IV fluids rather than restricted fluids are recommended for ABM in settings with high mortality and where patients present late. Cerebral Perfusion Pressure targeted fluid therapy (CPP>50 mm) is feasible and may improve outcome. Several newer anti-inflammatory agents are being investigated for treatment of ABM, and may hold promise for future management.

Session: MISCELLANEOUS II

BENIGN TRANSITIONAL DISORDERS AND THEIR DIFFERENTIAL DIAGNOSES

Andre Venter

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The majority of conditions that can be described as benign transitional disorders are often confused with epileptic seizures, and are therefore also referred as 'non-epileptic paroxysmal disorders'. Some conditions, although non epileptic may in fact be ominous signs of another neurological diagnosis, but this talk include conditions that are (fairly) benign only. Non-epileptic paroxysmal events (NEPE's) are divided into physiological or organic and psychogenic groups. Physiological NEPE's also include normal paroxysmal behaviours that are neither seizures nor disorders. These will form the focus of this talk. They occur at any age, but there highest incidence is seen in childhood, particularly in the first year of life, when suggesting a differential diagnosis can be very demanding.

NEPE's are usually abrupt, transient episodes of abnormal involuntary movement or impaired coordination of voluntary actions. Unfortunately 20–30% of patients with NEPE's are misdiagnosed as being epileptogenic and they are often treated for life.

Non-epileptic movement disorders in neonates and infants that may be misdiagnosed include primitive reflexes (Moro, ATNR), jitteriness, tonic reflex seizures, benign paroxysmal torticollis, rhythmic behavioural movements, self-gratification disorder, Sandifer syndrome, benign neonatal sleep myoclonus, among others. In older children these movement disorders are represented by tics, mannerisms, compulsions, stereotopies, nightmares and night terrors, shuddering attacks and crying-out spells (breath holding) among others.

The majority of these conditions can be accurately diagnosed by taking a good history and asking the parents/caregivers to imitate the 'attacks'. Video recall on mobile phones has facilitated the diagnostic process as the NEPE's may not always be present at the time of consultation. An accurate diagnosis precludes embarking on a battery of unnecessary and unhelpful special investigation and treatments.

ADHD: FROM NEUROBIOLOGICAL BASIS TO DIAGNOSIS AND TREATMENT

Nikolay Zavadenko

The Russian National Research Medical University named after N. I. Pirogov, Neurology, Neurosurgery and Medical Genetics Department of Pediatric Faculty, MOSCOW, Russian Federation

Attention Deficit Hyperactivity Disorder (ADHD) is a common neurobehavioral disorder affecting 3–7% of school age children. Comorbid disorders, including developmental language and motor coordination disorder, aggressive behaviors and emotional disorders, may be present in many children with ADHD, leading to poor adaptation abilities, problems at school education and difficulties with the selection of optimal treatment.

The phenotype of ADHD is determined by complex interaction of neurobiological and psycho-social factors. In our sample of 204 children (170 boys and 34 girls) aged from 5 to 13 years, who met ICD-10 and DSM-IV diagnostic criteria of ADHD, 84% of cases were suspected to be determined by early brain damage due to the pregnancy or delivery complications. Pedigree studies evidenced for genetic causality in 57% of patients, having first-degree relatives with ADHD. However, 42% of cases were caused by the combination of early brain damage and genetic factors. Intrafamilial psycho-social adversity seemed to play additional role and was revealed in 63% of cases.

The "Strengths and Difficulties" questionnaire [Goodman R., 1999, 2001] was put to the parents and teachers of 44 children with ADHD and 257 healthy schoolchildren aged 7–11 years. The spectrum of impairments arising in ADHD was found not to be restricted to the core symptoms of ADHD. Questionnaires completed by both parents and teachers showed that emotional impairments, behavioral problems, difficulties interacting with peers, and underdevelopment of social behavior were significantly more prominent in children with ADHD as compared with their peers. This study confirms the considerable negative impact of difficulties experienced by children with ADHD on their emotional state, family life, friendships, school achievements, and even leisure activities. The problems in school learning, behavior and emotional control can be related not only with ADHD itself, but also with the co-morbid disorders, which must be diagnosed properly and in time. The follow-up of the patients with ADHD must include systematic assessments of their social-psychological functioning, in accordance with the "Broader efficacy of treatment" concept.

EVALUATING THE OUTCOMES OF ATTENTION REHABILITATION IN CHILDREN WITH FOCAL EPILEPSY

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Aims. Few systematically controlled neurocognitive rehabilitation techniques for children exist. On the basis of FORAMENRehab software (originally by Sarajuuri et al, 2000) we conducted an intervention program for children with attention deficit.

Methods. 8 patients (M=10.43 y, SD=0.68) with focal epilepsy participated and received individual supervised computer-based training twice a week during 5-week-period using FORAMENRehab Attention module following a strict protocol. 3 difficulty levels were used in training. Before and after the intervention the children were tested with baseline assessment tasks.

Results. We present the development pattern and baseline performance (BP) comparison for four attention functions:

1) Focused: children reached 2nd level after 1.94 trainings (mean); all but one reached 3rd level after 4.21 trainings. Though, BP did not improve statistically. 2) Sustained: 2nd level attained after 1.8 trainings; all but one reached 3rd level after 3.37 trainings. BP: correct answers increased ($p < .02$). 3) Complex: In Paced Search task children stayed at the easiest level. BP: improvement in correct answers ($p < .02$). In Word Recognition task half of children reached 2nd level after 5 sessions, only one reached 3rd level. BP: improvement in correct choices ($p < .03$). In Addition task all but one reached 2nd level after 5.29 trainings, none reached 3rd level. BP: correct answers ($p = .042$) increased. 4) Tracking: development to higher levels not seen; BP: correct choices improved ($p < .05$).

Conclusions. The FORAMENRehab intervention is efficient for attention rehabilitation in children. Assessment of individual improvement showed that complicated tasks relate with slower progress. On the contrary, the greatest effects in baseline assessments were seen in complicated functions. Therefore, the efficacy of intervention can be seen through simultaneous analysis of individual improvement and improvement in baseline assessment tasks.

METHYLENETETRAHYDROFOLATE REDUCTASE POLYMORPHISMS C677T AND A1298C AS IMPORTANT GENETIC RISK FACTORS IN PEDIATRIC STROKE AND MIGRAINE

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Introduction. Methylene tetrahydrofolate reductase (MTHFR) impaired activity depending on common mutations (677C>T and 1298A>C) is considered a possible risk factor cerebrovascular events such as stroke and migraine. This study explores the role of the two MTHFR polymorphisms in Estonian pediatric stroke and migraine patients and to compare it with the population-based data.

Methods. A total of 81 pediatric patients (M=43, F=38) with a cerebrovascular disease (53 arterial ischemic stroke (AIS), 8 hemorrhagic stroke (HS) and 20 migraine) were included to the retrospective chart review and screened for common MTHFR mutations 677C>T and 1298A>C. Control group consisted of 763 (M=388, F=375) randomly selected healthy persons aged 18–30 years from the Biobank of Estonia. MTHFR polymorphisms were analyzed by polymerase chain reaction.

Results. The homozygous 677TT and the 1298CC genotypes showed a frequency of 9% and 8% in the stroke group and 10% and 10% in the migraine group, which were moderately higher compared with the general population (7% and 6% respectively). A higher frequency of the C allele was found in locus 1298 in the migraine group (33%) compared with the general population (28%). Of the combined genotypes, the highest prevalence was represented by the 677CC/1298AC combination. The average serum homocystein level was significantly higher in the migraine group than in the AIS and HS groups, but mean values were in the normal range.

Conclusion. A higher prevalence of mutations causing impaired MTHFR activity was found in patients, which was more pronounced in the migraine group. This association demands further larger-scale studies as folate-pathway activity enhancing agents could be used to limit or prevent the disease in the genetically predisposed population. A screening for risk and recurrence evaluation and treatment including a consideration of a nutrition policy based on folic acid supplementation could be introduced if a positive association existed.

POSTER PRESENTATIONS

May 30, Thursday

RARE DISORDERS: AUTOIMMUNE, NEUROMUSCULAR, METABOLIC/GENETIC

PEDIATRIC ONSET MULTIPLE SCLEROSIS: THE DATA OF PEDIATRIC NEUROLOGY UNIT IN KAUNAS

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Objective. To evaluate the clinical characteristics, diagnostic and treatment data of pediatric onset multiple sclerosis (MS).

Methods. A retrospective cohort included data of 32 patients diagnosed with MS before the age of 18 years evaluated during the period of years 2000–2012 in Pediatric Unit of Neurology Department of Hospital of Lithuanian University of Health Sciences.

Results. Twenty patients (63.3%) were girls and 12 were boys (36.7%), the sex ratio female/male was 1.6: 1. Age at onset of MS ranged from 7 to 18 years (median 14.5). In 7 out of 32 patients disease started before 12 years of age, and in 25 children (78.1%) after. The most frequent first symptom at disease manifestation was sensory (50.0%), brainstem (40.6%) and motor (31.3%), optic neuritis occurred in 18.8% of cases. All patients presented with relapsing-remitting course of disease. In 17 (54.8%) patients relapse occurred during the first year after MS diagnosis was established with a mean relapse rate of 2 ± 1.1 . Seven patients experienced 3 or more relapses within that period of time. Head MRI data were available for all patients whereas spinal MRI was performed only for 12 (37.5%) patients with pathological findings in all cases. Visual evoked potentials were examined in 25 children with pathologic findings in 76.0%, brainstem auditory evoked potentials in 29 patients with abnormal findings in 44.8% of cases. A total 22 (68.75%) patients were investigated for oligoclonal bands in cerebrospinal fluid. Out of them in 9/22 (40.9%) cases oligoclonal bands were found to be positive, in 18.2% negative and in 40.9% results were not available. Cognitive and psychological profile studies were not performed. EDSS mean score was 2.23 ± 1.1 at the moment of MS diagnosis, and 1.27 ± 0.6 after prednisone i/v for 5 days. Immunomodulatory therapy became possible in 2010, since then 8 patients received treatment (1 with glatiramer acetate, 6 with interferon-beta and one with intravenous immunoglobulin).

Conclusion. To ensure evaluation and management of children and teenagers with MS standardized protocols and treatment guidelines are needed. Participation in MS registry, establishment of reference center could provide a comprehensive care and smooth transition to adulthood for this group of patients.

DIAGNOSING PEDIATRIC MULTIPLE SCLEROSIS ACCORDING TO BASELINE AND FOLLOW-UP CLINICAL AND MRI FINDINGS: COMPARISON OF REVISED CANADIAN AND MCDONALD MRI CRITERIA. THE PILOT-STUDY IN ESTONIA

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Background. Diagnosing multiple sclerosis (MS) in children and adolescents is still challenging. Canadian criteria (Callen, 2008) for using MRI findings to diagnose pediatric MS and a revised McDonald criteria (2010) are used. Canadian and McDonald criteria differ in numbers and in locations of brain lesions.

Objective. The aim of the pilot-study is to analyze MS clinical and radiological symptoms, follow-up MRI findings while comparing Canadian and McDonald MRI criteria.

Patients and materials. The study was carried out at the Children's Clinic of Tartu University Hospital, Tallinn Children's Hospital and West-Tallinn Central Hospital. The patients were retrospectively selected from the hospital database with MS diagnoses (2008 to 2012). Hospital records and MRI images were reviewed. 11 patients with clinically definite MS were included (8 girls and 3 boys). Mean age at the MS onset was 15.3 yr. All patients had a relapsing-remitting course. Follow-up brain MRI scan was performed on average 6 months after the first demyelinating attack on 8 patients. On 4 of them spinal MRI was also performed. During a one-year period (range 1.25–3.33 yr.) 6/8 patients experienced at least one relapse.

Results. The most common clinical symptoms of MS were the following: blurring of vision (6/8); problems with mobility and balance (6/8); numbness and fatigue – both (4/8). At the end of follow-up only 3 patients were symptomatic.

MRI evidence for DIS and/or DIT were used. The overall sensitivity of revised McDonald criteria to diagnose MS was 75% (6/8) and for Canadian criteria 100% (8/8). There were no new T1 hypointense lesions found in follow-up studies, while 7/8 patients had over 20 T2 or T2/FLAIR hyperintense lesions. Brainstem and/or spinal cord lesions were detected in 3 cases on initial MRI and in 5 cases on follow-up.

Conclusions. Canadian criteria to diagnose pediatric MS seems to be more sensitive than revised McDonald criteria.

ANTI-N-METHYLSPARTATE RECEPTOR ENCEPHALITIS IN A YOUNG CHILD: A CASE REPORT

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Introduction. Anti-N-methylaspartate receptor (NMDAR) encephalitis is a recently identified synaptic autoimmune disorder that is increasingly recognized. It can occur at any age with children representing about 40% of all cases.

Case. A previously healthy 2 years old boy had a fever one week before the first neurological symptoms. He presented

with febrile epileptic seizures evolving to status epilepticus. At the moment his neurological exam, imaging and cerebrospinal fluid were normal. Within next 2 weeks he remained irritable, lost eye contact and interest to surrounding, developed tremor of the body. Subsequently he had repeated partial seizures, lost motor skills, became somnolent. In the next few days he developed orofacial dyskinesias and hyperkinetic movements of the limbs and became comatose. He remained in Pediatric Intensive Care Unit for 23 days intubated, in a violent restlessness and hyperkinetic state expressed by choreiform movements, orofacial dyskinesias, constant movements of the tongue. He had resistant hyperthermia and tachycardia. Brain MRI was normal, work-up to exclude neurotropic viruses, intoxication, metabolic disease, vasculitis was negative. Treatment included antiepileptic drugs, sedatives, neuroleptics, steroids and IVIG without response. In the following 3 months he remained unresponsive, in constant hyperkinetic movements of the face and body, repetitive seizures, severe sleep disruption, hyperthermia. Similar although less severe pattern continued during the next year. After 1 year 9 months from the disease onset his diagnosis was reconsidered, serum was tested at the John Radcliffe Hospital, Oxford, UK for antibodies against anti-NMDA receptors and found to be positive. He remains with considerable residual deficit at the latest follow up 3 years after disease onset: walks independently but purposelessly, he doesn't speak and has no social contact. He sleeps well, has no hyperkinetic movements or seizures although EEG shows multifocal epileptiform discharges.

Conclusion. Diagnosis of NMDAR encephalitis might be the diagnostic challenge in a very young age. Recognition of special combination of neurological signs with multistage development should prompt the suspicion of NMDAR encephalitis followed by testing of specific antibodies and appropriate treatment.

ANTIBODY-NEGATIVE AUTOIMMUNE ENCEPHALITIS IN A 4-YEAR-OLD ESTONIAN BOY

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Background. Autoimmune encephalitis (AIE) is rare in children and usually occurs after viral infection. Patients typically present with seizures, memory problems, and imaging changes in the medial temporal lobes.

Clinical case. A 4-year old boy was admitted due to progressive somnolence, drooling, inability to swallow, difficulties to talk and walk during the past 3 days after respiratory infection. Before that he's been a healthy boy. On clinical examination he was somnolent, had tetraparesis, marked hypotonia, no pathological reflexes, drooling, hypomimic face and no verbal communication. All routine laboratory tests were normal. Cerebrospinal fluid (CSF) analysis was within normal range. All test for infectious etiology came back negative. Autoantibodies (a-Ab) for AIE in CSF and serum (NMDA, GABA(b), AMPA 1, LGI 1, AMPA 2, Caspr 2) were negative. Typical a-Ab in a rheumatic panel was also negative, except Jo1 IgG titer that was 5-fold elevated. Full metabolic workup was negative. Genetic testing for Wilson disease was negative. Brain MRI scans were performed on the 4th, 10th, 17th day of disease. 1st MRI showed limbic encephalitis and bilateral

hyperintensive lesions of cortex, 2nd MRI revealed lesions of basal ganglia but lesions of cortex had subsided. Last MRI was without any improvement. Electroencephalogram (EEG) showed bilateral synchronous slowing without any epileptic discharges. Electroneuromyography was normal. A diagnostic search for cancer with whole body CT was negative. Treatment with intravenous acyclovir was started immediately and continued for 21 days. After suspecting an autoimmune etiology immunomodulatory treatment with i/v immunoglobulin, oral and pulse therapy with glucocorticoids was initiated, along with 4 plasmaphereses that were performed. With treatment the child has no impairment of consciousness: a week after admission the child follows with eyes, reacts with emotions quite adequately, but still no verbal contact. 2 weeks after admission spasticity and extrapyramidal signs developed. 5 weeks after admission a clinically child improves: still some extrapyramidal signs, but no hyperkinesia any more.

Conclusion. The diagnosis of AIE is very challenging in children, but they still benefit from immunomodulatory treatment.

FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY. CASE REPORT

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Introduction. Facioscapulohumeral muscular dystrophy (FSHD), also known as Landouzy-Dejerine, is a usually autosomal dominant inherited form of muscular dystrophy (MD) that initially affects the skeletal muscles of the face, scapula and upper arms. While it is widely stated to be the third most common genetic disease of skeletal muscle, a 2008 analysis of rare diseases listed FSHD as the most prevalent form of MD at 7/100000.

Symptoms may develop in early childhood and are usually noticeable in the teenage years with 95% of affected individuals manifesting disease by age 20 years. A progressive skeletal muscle weakness usually develops in other areas of the body as well; often the weakness is asymmetrical. Non-muscular symptoms frequently associated with FSHD include subclinical sensorineural hearing loss and retinal telangiectasia. In more than 95% of known cases, the disease is associated with deletions of the D4Z4 repeat in the 4q35 subtelomeric region of Chromosome 4.

Case. We report on a 15-year old Estonian male patient who was referred to our EMG laboratory with foot drop on his right leg, for confirmation whether it is peroneal nerve neuropathy. Physical examination showed also asymmetrical facial and shoulder girdle muscle weakness. EMG: myopathic pattern, asymmetrical, on various stages in different muscles. Normal nerve conduction velocities. Serum CK mildly elevated 901 U/l. Muscle biopsy (tibial anterior muscle) reveals myopathic changes, most possibly limb girdle MD. DNA analysis confirmed the deletion of chromosome 4q35.

Conclusion. FSHD is a distinct type of muscular dystrophy, both clinically and genetically. Being one of the most prevalence forms of MD it tends to be under-diagnosed. With our report we would like to present you the early signs, clinical evaluation and diagnostics of FSHD.

POLYNEUROPATHY IN CHILDREN WITH INBORN ERRORS OF METABOLISM

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The peripheral nervous system (PNS) can be involved into the pathology in patients with inborn errors of metabolism (IEM). In most cases, polyneuropathy is part of a diffuse clinical picture. Review of IEMs causing neuropathies in children and case reports of long-term treated patients are presented. Acute polyneuropathy (AP) is characteristic for hereditary tyrosinemia type 1 (HT1), acute intermittent porphyria (rarely before puberty), pyruvate dehydrogenase deficiency (PDD). Chronic (predominantly) axonal polyneuropathy (CAP) – for mitochondrial trifunctional proteina, isolated long chain acyl-CoA dehydrogenase (LCHAD), biotinidase, vitamin E, serine, ornithine aminotransferase deficiencies, homocysteine remethylation defects, PDD, Wilson disease, cerebrotendinous xanthomatosis. Chronic (predominantly) demyelinating polyneuropathy (CDP) – for lysosomal storage diseases involving myelin metabolism, peroxisomal diseases, respiratory chain disorders.

Case 1: HT1 patient, 16y old boy, suffered neurological crisis (NC) with AP before the diagnosis of the disease and after interruption of nitisinone (NTBC) treatment at age 3 y. After re-introduction of NTBC the patient regained neurological functions very slowly, significant long-lasting ENMG alterations (axonal degeneration with secondary demyelination).

Case 2: HT1 patient, 11y old girl, received an uninterrupted treatment with NTBC since age 2y after the diagnosis of the disease. Still avoids the development of PNS damage.

Case 3: patient with LCHAD deficiency, 14y old girl, suffered Reye syndrome-like episodes since age 10mo, recurrent episodes of rhabdomyolysis since age 3y. Reversible ENMG symptoms of polyneuropathy during long-term treatment.

Conclusions. AP and CAP are characteristic for treatable IEMs. Appropriate diagnostics and management of treatable IEMs can prevent PNS damage and development of irreversible clinical dysfunction and ENMG alterations.

COMPREHENSIVE CARE OF TUBEROUS SCLEROSIS COMPLEX: ARE THE NEEDS MET?

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Background. Tuberous sclerosis complex (TSC) is a rare multisystem genetic disorder presenting with multiple hamartomatous lesions in different organs, sometimes causing critical states. Careful diagnostic evaluation according to internationally approved clinical criteria and follow-up of patients with TSC may help to avoid dangerous complications.

Objective. To analyze clinical manifestations in the cohort of patients with TSC and to assess the adherence to surveillance guidelines.

Methods. Retrospective analysis of medical records of all TSC cases registered within 1982–2012 at the Child Neurology and Child Neurosurgery Units, Hospital of Lithuanian Univer-

sity of Health Sciences. Surveillance adherence was recorded at diagnosis and during subsequent evaluations.

Results. The diagnosis of TSC was confirmed in 44 children. 93% of patients had at least one seizure in the course of the disease (refractory in 86%), 18% of them experienced epileptic status. Developmental delay was documented in 73% of patients. Brain CT or MRI was abnormal in all patients: tubers were detected in 83%, subependymal nodules in 81%, subependymal giant cell astrocytoma in 26% of cases (surgery performed in 4 of them). Renal angiomyolipomas were found in 22% of cases (renal bleeding developed in 2 cases), cardiac rhabdomyoma in 32%, hepatic tumors in 13%, retinal hamartomas in 21%, cutaneous symptoms in all patients.

Initial evaluation was completely adequate to surveillance guidelines in 26% cases. Follow up evaluation was inadequate in 58% of patients, more than one diagnostic procedure was lacking in 35% of children.

Conclusion. Our patients had variable clinical manifestations of TSC, at least ¼ developed life-threatening complications. In order to ensure appropriate care of patients with TSC, careful adherence to standardized protocols and treatment guidelines are needed. Involvement in the TSC registry and the activities of the newly established competence center for phacomatosis in Kaunas may help to provide better comprehensive service for this group of patients.

NEUROFIBROMATOSIS TYPE 1: MANAGEMENT AND NEEDS IN LITHUANIA

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Background. Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder with variable expression. Neurologic complications include tumors of the brain and of the peripheral nerves, nerve roots, and plexi; spinal cord compression; dural ectasias; learning disabilities; attention deficit; headaches; seizures; deafness; hydrocephalus and stroke. For this group of patients, careful follow-up and early intervention in cases with tumor progression is very important.

Objectives. To assess the prevalence, clinical symptoms and management of children with NF1.

Methods. We retrospectively analyzed case histories of all patients followed by the paediatricians, neurosurgeons and child neurologists at the Hospital of Lithuanian University of Health Sciences Kaunas Clinics.

Results. Since 2007, the diagnosis of NF1 was confirmed in 32 cases, 17 (53.1%) boys, 15 (46.9%) girls, confirmed by biopsy in 13 (40.6%) cases. Family history of NF1 was recorded in 9 (28.1%) cases. As initial symptoms, *café au lait* spots since birth were recorded in 28 (87.5%) cases, ophthalmological problems in 25 (78.1%), neurofibroma in 1 case, backpain in 1 case, insufficiency of breathing (because of mediastinum tumor) in 1 case. During the follow-up, freckling of the axillae or inguinal regions were documented in 8 (25%) cases, dermal neurofibromas in 18 (56.2%), plexiform neurofibromas in 5 (15.6%), Lisch nodules in 5 (15.6%), orthopedic problems in 6 (18.7%), endocrinological problems in 10 (31.2%), learning disabilities and attention deficit in 11 (34.4%), pain in 9 (28.1%) cases, strabismus in 10 (31.2%), impairment of visual acuity in 21 (35.6%) cases. Optic tract glioma was diagnosed in 17 (53.1%) cases, spinal tumours in 2 (6.2%), tumours in other organs in 13 (40.6%), epilepsy in 2 (6.2%) cases. Brain surgery was performed in 4 (12.5%), shunting

due to hydrocephalus in 3 cases. Chemotherapy was given to 18 (56.25%) patients, the mean number of hospitalizations for that was 134.

Conclusion. This pilot survey showed that just around 20% of possible NF1 cases are under careful follow-up, mainly due to neural tumors. In most cases, vision problem was the first sign leading for diagnostic work-up, despite of the fact that *café au lait* spots were present since birth. The diagnosis of NF1 was delayed in many cases, possibly due to insufficient awareness of medical professionals. Elaboration of management guidelines, a coordinated multidisciplinary approach and follow-up at the specialized center, also education of medical specialists and families about the possible symptoms and management strategies may help to avoid severe consequences of this familial disorder both in childhood and later in adulthood.

SMALL SUPERNUMERARY MARKER CHROMOSOMES DERIVED FROM CHROMOSOME 15

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Introduction. Small supernumerary marker chromosomes (SMC) are an extra structurally abnormal chromosomes that cannot be identified or characterized by conventional cytogenetics techniques alone. Approximately 30% of SMC are derived from chromosome 15 (SMC(15)). The proximal region of chromosome 15 long arm is unstable and predisposed to genomic rearrangements. The genes in 15q11-q13 region undergo to parental imprinting also, and deletions of this region leads to Prader-Willi or Angelman syndrome.

The aim of study is to identify and characterize SMC, which was detected in standard karyotype analysis.

Materials and Methods. The SMC were characterized by fluorescent in situ hybridization method (FISH) with probe for Prader-Willi/Angelman syndrome critical region (PWACR or SNRPN locus) from Abbott Molecular, Inc. in three children with various health problems.

Results. A newborn girl with skin problems resembling incontinentia pigmenta identified non mosaic SMC contained two D15Z1 loci and not PWACR, and the karyotype was interpreted as 47,XX,+idic(15)(q11.2).ish idic(15)(q11.2)(D15Z1++,SNRPN-). A 5-year-old girl with psychomotor development delay and minor anomalies had identified SMC, which was derived from chromosome 15 and contained two D15Z1 and two SNRPN loci: 47,XX,+mar.ish der(15)(D15Z1++,SNRPN++). A 4-year-old girl with muscular hypotonia, psychomotor development delay, facial dysmorphism, chest deformity had identified mosaic SMC in 90% of analysed cells, which was characterized as idic(15)(q12)(D15Z1++,SNRPN++).

Conclusions. We demonstrate three children with different SMC derived from chromosome 15. Two patients had inverted duplication of chromosome 15 with breakpoints at 15q11.2 and 15q12 respectively, and one patient had other structural rearrangement of derivative chromosome 15. Two patients had tetrasomy of PWACR and 15p, and one patient had only tetrasomy of 15p.

CASE REPORT OF TWO NEWBORN BOY WITH CONGENITAL ANOMALIES AND RING CHROMOSOMES

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Introduction. Ring chromosomes are rare finding that can be reason of congenital anomalies and mental retardation.

Case 1. A newborn boy had coarse facial features, bilateral epicanthal folds, hyperthelorism, umbilical hernia, swollen feet, deformation of the second toes of both feet, scrotal pigmentation and micropenis. The boy had atrial septal defect and peripheral pulmonary stenosis. Conventional cytogenetic analysis on peripheral lymphocytes revealed that one chromosome 18 had formed a shape of ring. The fluorescent in situ hybridization (FISH) studies showed the deletion of both subtelomeric regions of ring chromosome 18, and karyotype was interpreted as 46,XY,r(18)(p11.3q23).ish r(18)(D18S552-,D18S1390-).

Case 2. A newborn boy was consulted due to intruterine growth retardation, partial intestinal obstruction with meconium, rethinopatia congenita of the right eye and leukocornea of the left eye. Karyotype analysis on peripheral lymphocytes was performed that revealed mosaicism of two cell lines – 46,X,r(Y)(p11.32q11.2~12)[10]/45,X[20].

Conclusions. We demonstrate two rare constitutional chromosome aberrations formed by ring shaped chromosomes which depending on involved chromosomes can cause different clinical manifestation.

The first case is a newborn boy with ring chromosome 18 who presents dysmorphic features, congenital heart disease, umbilical hernia and micropenis. These clinical findings have been described in other similar cases of ring chromosome 18.

The second case is about newborn boy who was born premature, with small for gestational age and with partial intestinal obstruction and leukocornea. His karyotype revealed mosaicism of cell line with 45,X and the other cell line with ring chromosome Y. Based on literature data part of his clinical findings may be caused by his chromosomal aberrations.

SUBTELOMERIC FISH ANALYSIS IN NEWBORNS WITH CONGENITAL ANOMALIES

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Introduction. The prevalence of congenital anomalies are estimated about 3–5% in newborns. The congenital anomalies can be caused by many factors, including genetic ones. The recent studies demonstrate the role of submicroscopic chromosomal rearrangements in etiology of congenital anomalies.

The aim of the study was to reveal and characterise subtelomeric chromosome aberrations in a cohort of Latvian newborns with congenital anomalies.

Materials and Methods. We examined 32 newborns with congenital anomalies and normal standard karyotype by ToTelVysion Multi-color fluorescent in situ hybridization (FISH) probe set (Abbott Molecular, Inc.).

Results. The subtelomeric chromosomal rearrangements were detected in 4 (12.5%) patients. Three patients had deletions (1pter, 3pter, 6qter), and one patient had unbalanced translocation der(1)t(1;2)(pter-;pter+) inherited from father. All patients had intrauterine growth retardation, premature birth, microcephaly, congenital heart disease, corpus callosum hypoplasia or agenesis, facial dysmorphism, overlapping fingers or toes.

Conclusions. Our results show the importance to screen for submicroscopic subtelomeric chromosomal abnormalities in newborns with unexplained spectrum of various clinical symptoms.

JUVENILE HUNTINGTON DISEASE: CASE REPORT

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Huntington disease is an autosomal-dominant disorder with onset in midlife caused by an excess of CAG nucleotide repeats in the HTT gene, coding for huntingtin protein, and is classically characterized by motor, cognitive, and psychiatric disturbances. Juvenile forms of Huntington's disease is far less common and may have presentation with different motor signs including parkinsonism, dystonia and ataxia.

We present a 13 years old boy with Juvenile Huntington disease, with probable maternally inherited transmission. The patient was the second child of a non-consanguineous healthy Lithuanian couple with a normal birth and early development stages. The family is socially challenged as the mother has been a drinker and left the family when the boy was at preschool age. His father, aged 43 years, has no evidence of neurological disease. The patient's maternal grandmother had clinical symptoms of Huntington disease. When the boy was 8 years old learning difficulties appeared. At the age of 12 years, the patient started presenting articulatory disturbances and gait difficulties. His condition deteriorated gradually and by the age of 13 he had developed mild cognitive impairment, choreiform movements in the extremities, trunk, and face, dystonic arm posturing, and bradykinesia. There was no evidence of seizures or myoclonus. He had attended special education classes since primary school graduation. Neuropsychological testing revealed a low IQ (WISC score = 72).

The (CAG)_n triplet repeat of HTT gene was determined using quantitative fluorescent PCR with specific primers and fragment length analysis. The genotype of the patient is (CAG)_n=14/56. Full penetrance HD-causing allele ((CAG)_n=56) was identified.

Our patient showed all typical symptoms of Huntington's disease which was confirmed by molecular genetic analysis. We conclude that Juvenile Huntington disease should be considered in children suffering from a progressive neurodegenerative disease. It is important to be aware of hereditary conditions such as Huntington disease and to provide family counselling before genetic testing and after the diagnosis is confirmed.

May 31, Saturday

EPILEPSY. DEVELOPMENT. MISCELLANEOUS

THE FIRST EXPERIENCE WITH LEVETIRACETAM THERAPY IN REFRACTORY CHILDHOOD EPILEPSY

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Introduction. Levetiracetam (LEV) is widely used in the treatment of refractory childhood epilepsy, though in Lithuania it is one of the recently available drugs. Clinical studies have confirmed its efficacy in both generalized and focal epilepsy with minimal adverse effects.

The aim of study. To evaluate an efficacy and tolerability of LEV as an add-on therapy in children with refractory epilepsy.

Methods. Retrospective review of medical records of children with refractory epilepsy followed at the Hospital of Lithuanian University of Health Sciences in Kaunas within October 2009 – February, 2013.

Results. LEV as add-on therapy was introduced in 48 children (age range 1–17 years, mean age 9 ± 4.7), 56% of them suffering from refractory focal cryptogenic/symptomatic epilepsy (including 27% with ESES-like EEG changes), 29% having generalized symptomatic, 12% – generalized idiopathic epilepsy, and 4% – Dravet syndrome. Concomitant developmental disorders (mental retardation, behavioral, speech or motor disorder) prior to LEV treatment were found in 88% of cases. Most patients were on polytherapy (mean number of antiepileptic drugs used before LEV was 4.9 ± 2), 54% had undergone steroid therapy. Mean maximum dose of LEV was 1302 ± 683 mg/d, duration of treatment 14.8 ± 14 months. Treatment improved seizure control and was therefore continued in 27/48 (56%) cases. The benefits from LEV were observed in cryptogenic/symptomatic epilepsy with (64%) or without (53%) ESES-like changes on EEG, generalized symptomatic epilepsy (57%), also Dravet syndrome. In 16/48 (33%) improvement of mental or behavioral status has been documented. LEV was withdrawn in 21/48 (44%) cases within 5 ± 4.7 months due to poor efficacy or tolerability: in 12/21 cases due to ineffectiveness, and in 9/21 cases due to adverse effects. Adverse effects were noted in 23/48 (48%) cases: behavioral/emotional disorders in 13/48 (27%, 5 of them had to withdraw LEV due to this reason), somnolence in 4, nausea in 2, speech regression in 1 case; 21 out of 23 patients who experienced adverse effects had concomitant developmental disorder prior to LEV treatment.

Conclusion. LEV proved to be sufficiently (>50%) and comparatively effective in different types of drug-resistant childhood epilepsies. Adverse effects were more common in patients with developmental disorders, mainly the behavioural ones.

TREATMENT OF REFRACTORY EPILEPSY WITH METHYLPREDNISOLONE IN A PATIENT WITH GAUCHER DISEASE (NEURONOPATHIC PHENOTYPE). A CASE REPORT

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Background. Gaucher's disease is a lysosomal storage disease caused by the lack of beta-glucocerebrosidase enzyme, leading to the accumulation of glucocerebroside.

Clinically, two forms of Gaucher's disease are defined: nonneuronopathic form, so-called type 1, characterized by hepatosplenomegaly, thrombocytopenia, anemia, and osteopenia, and neuronopathic form, known as types 2 and 3, which are also characterized by hepatosplenomegaly, hematological and bone changes; however, involvement of the central nervous system dominates in the clinical picture (oculomotor apraxia, ataxia, mental retardation, bulbar symptoms, spasticity and progressive myoclonus epilepsy).

Case report. A full-term girl (A.D.) from the first uncomplicated pregnancy firstly presented with splenomegaly, hepatomegaly, anemia and thrombocytopenia. Besides oculomotor apraxia, ataxia, slight delay in psychomotor development were documented. Molecular examination revealed the genotype C721G>/C1448T>C (neuronopathic phenotype). Enzymatic replacement therapy with imiglucerase at a dosage of 60 U/kg every 2 weeks was administered. During half a year of the therapy, normalization of hematological parameters was achieved together with normalization of spleen and liver volumes. Neurological symptoms persist with myoclonic, generalized tonic-clonic seizures refractory to antiepileptic drugs. Intravenous methylprednisolone 20 mg/kg/d was given for 5 days in addition to a stable dosage of the regular antiepileptic drugs. The seizure frequency decreased markedly. There were no complications associated with intravenous methylprednisolone.

Conclusion. Methylprednisolone pulse therapy might be an option for the treatment of refractory seizures in children with Gaucher disease.

LINEAR SCLERODERMA EN COUP DE SABRE ASSOCIATED WITH EPILEPTIC SEIZURES: A CASE REPORT

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Introduction. Linear scleroderma is a rare form of localized scleroderma that mostly affects pediatric population. When linear scleroderma occurs on the head, it is referred to as linear scleroderma en coup de sabre (LSCs). Almost one-fourth of children with LSCs may develop extracutaneous manifestation. Nervous system involvement includes seizures, headache, focal neurologic deficits and neuropsychiatric symptoms.

Case. 4-years-old previously healthy girl developed first episode of febrile seizures. On the next day skin lesion on left side of forehead has appeared. During the next six months skin lesion has progressed with the development of ivory colored sclerotic plaque on the left forehead extending to medial angle of the eye and mouth as well as alopecia. She was diag-

nosed with linear scleroderma and received topical treatment. At the same time she has experienced 2 non provoked focal seizures. EEG showed a single spike-wave pattern in the left frontal area and treatment with valproates was initiated. Physical examination showed normal neurological status and other systemic findings were unremarkable. Complete blood count, erythrocyte sedimentation rate and biochemical findings were normal. Rheumatoid factor, antinuclear antibodies and antibodies for chronic infections (cytomegalovirus, Epstein-Barr virus) were negative. Head CT showed no abnormalities. Head MRI showed ipsilateral T2 hyperintensities in periventricular region.

Conclusion. Neurological manifestation can precede or occur concurrent with the skin lesion in patients with linear scleroderma. The relation between LSCs and seizures can be due to local structural CNS lesions which are most likely related to perivascular infiltrate and vasculitis possibly leading to chronic inflammatory process whereas the presence of neurological symptoms and radiologic abnormalities requires close clinical follow-up.

DRAVET SYNDROME (A CASE REPORT)

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Dravet syndrome (DS) is a rare form of intractable epilepsy. Patient with DS diagnosed at the age of 1y is presented. The girls's perinatal anamnesis was normal. The mother's aunt has epilepsy. Normal development before onset of seizures. At the age of 3.5mo: 2 weeks after vaccination focal epilepsy seizures started (clonic convulsions of left hand) re-occurring up to 5 times per day for 1 week. Left-side clonic convulsion seizure (duration 20 min) presented after a hot bath at the age of 4 mo. Long-term sleep video EEG was without evident epileptic activity (EA). Brain MRI: bilateral frontal lobe atrophy. No treatment prescribed. Age 7mo: left-side clonic convulsions with generalization (duration 25 min) re-occurred during infection. Sleep EEG: local EA in CT area. Age 7.5mo: 5 left-side clonic convulsions with generalization re-occurred during infectious diseases and subfebrile temperature. 2 of them continued until status epilepticus (SE). Age 9mo: right-side afebrile clonic convulsions re-occurred. Febrile or afebrile alternating hemi-convulsions – frequent, tendency of re-occurrence at the same day continuing until SE. Todd paresis was observed after seizures. Age 1y: eyelid myoclonia started with multiple re-occurrences during the day. Treatment before DS diagnosis – OXC, CBZ, PB, CNZ, with no effect. DS was diagnosed at the age of 1y. Treatment was changed: VPA, TPM, CLZ. Hemi-convulsions with generalization became less frequent, myoclonic seizures persisted. Age 1y3mo: atypical absences, eyelid myoclonia re-occurring many times per day occurred. Presently age 3.5y: eyelid myoclonia, atypical absences became less frequent, rare hemi-convulsions evolving to SE (provoked by infection and hyperthermia), EEG: multiple local and generalized changes of EA; disorder of cognitive functions, speech prevail, behavioural disorder, independent walking (since 1y8mo), ataxia.

Conclusion. Early recognition of DS is important for efficiency of the treatment and disease prognosis.

CDKL5 GENE RELATED EPILEPTIC ENCEPHALOPATHY IN ESTONIA: A CASE PRESENTATION

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Background. X-linked aberrations of Cyclin-dependent-kinase like 5 (CDKL5) gene in female patients cause a rare form of an early-onset epileptic encephalopathy: intractable seizures, developmental delay and Rett syndrome-like features.

Clinical case. A 2 months old girl was referred to pediatric neurologist due to epileptic seizures described as series of spasms. Before that she's been a healthy child. Neurologic examination showed poor psychomotor development: lack of eye contact and also severe hypotonia. Despite treatment with valproic acid, seizures continued. At 4 months she had poor head control, lack of emotional contact and persistent hypotonia. She presented spasms and tonic-clonic seizures 2–10 times per day. Electroencephalography (EEG) showed some focal origin and secondary generalized epileptic discharges from right hemisphere with a normal baseline activity. Magnetic resonance tomography (MRT) showed no structural abnormalities. At the age of 6 months she developed myoclonic seizures. Several antiepileptic drugs (AED's) and combinations of drugs were used to treat the seizures but none was effective. EEG at this point showed bilateral paroxysms of spikes and slow waves. Different genetic tests were performed: metabolic workup, POLG gene mutation, GLUT1 mutation, all negative. MRT and MRS (magnetic resonance spectroscopy) were done repeatedly without any abnormalities. Electroneuromyography, ECG, US of heart and other organs were normal. Treatment with ketogenic diet was tried but with no effect. In CDKL5 gene sequence analyze a heterozygous mutation c.1648C>T (p.R550X) was found in exon 12 of CDKL5 gene and a diagnose of an autosomal dominant X-linked epileptic encephalopathy (atypical Rett syndrome) was made. At the age of 1.5 years the girl had a severe developmental delay: highest motor function was rolling over both sides, brief eye contact was present but no smiling or active communication. Severely decreased muscle tone persisted. She had no difficulties with swallowing or eating. Myoclonic seizures were present up to 10 times a day. No effect of any AED's was achieved.

Conclusion. This was the very first case of CDKL5 epileptic encephalopathy in Estonia. We suggest that genetic testing of CDKL5 should be considered in girls with early-onset epileptic encephalopathy and RTT-like features.

LATE ONSET SPASMS IN A BOY WITH RHIZOMELIC CHONDRODYSPLASIA PUNCTATA

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Background. Late-onset spasms (LOS) are epileptic spasms starting after the first year of life. Few reports have been published on LOS after infancy period. They are more frequently observed in patients with epileptic encephalopathy and not commonly accompanied by hypsarrhythmia.

Case report. We present a 7 year old boy with rhizomelic chondrodysplasia punctata (RCDP) type 2 who developed clusters of flexion type spasms at the age of 6 years. He is the 1st and only child of first degree consanguineous parents.

Family history is unremarkable. He was born at term (birth weight 2.8 kg) after uneventful pregnancy by CS due to oligohydramnion (found to have hydronephrosis). His early developmental period was marked by failure to thrive and delayed motor and mental development. He is fed by GT and wheel – chair bound. He underwent eye surgery due to congenital cataract. He is grossly growth delayed with marked shortening of his proximal limbs, disproportionate dwarfism and multiple joint contractures. He is microcephalic with dysmorphic features: large ears, elongated philtrum, upturned nares, small phallus, undescended testes. Comprehensive peroxysomal screen revealed almost absent dihydroxyacetonephosphate acyltransferase (DHAPAT) activity (1st enzyme in plasmalogen synthesis). Also deficient de novo plasmalogen synthesis in his fibroblasts was found, while his very long chain fatty acid profile was normal. Brain MRI at the age of 3 years showed delayed normal myelination, subtle decreased bulk of posterior cranial fossa, vertebral clefts C7/T6 compatible with RCDP. The result of molecular-genetic test has not been available. Interictal EEG showed hypsarrhythmic pattern (disorganized BGA, multifocal and generalized epileptiform bursts). He was immediately started on vigabatrin and responded well, without adverse drug effects and have been staying on it for 9 months. Spasms were gradually disappeared and EEG has been improved. On last EEG, BGA was normalized while bilateral pronounced focal fronto-temporal epileptiform changes remained.

Conclusions. Late onset epileptic spasms accompanied by hypsarrhythmia can be one of presenting feature of epilepsy in patients with RCPD, most likely corresponding to dysfunctional and delayed maturational process. The clinical and neurophysiological response on vigabatrin has been favorable.

PERCEPTIONS AND ATTITUDES OF ADOLESCENTS TOWARDS THEIR EPILEPSY

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Introduction. The quality of life of a child with epilepsy may be affected by the constraints in daily life due to safety issues and medical follow up procedures even in cases when epilepsy is well controlled.

Objectives. To investigate the attitudes of adolescents towards their disease and their subjective perceptions of the impact that epilepsy has on their quality of life.

Subjects and methods. We studied patients aged 12–17 years who had the diagnosis of epilepsy and were on treatment with antiepileptic drugs for more than one year. The study cases were recruited from the inpatients at the Child Neurology unit, the Hospital of Lithuanian University of Health Sciences Kauno klinikos, from 2009 to 2010. All patients filled out the questionnaire with questions about their perceptions of the medical and social aspects of their epilepsy. The objective medical data were supplemented by the relevant hospital documentation.

Results. A total of 71 children were enrolled in this study. In 54.9% of cases the course of epilepsy was self-reported as easy-going, in 26.8% as moderate, in 22.5% as severe one. Only 28.2% of the respondents were anxious about their illness, while 71.8% others reported that the disease did not bother them too much. Regarding the impact of schooling, 49.3% of respondents have never skipped the classes because of their disease, 46.5% did that occasionally, and 4.2% often. The absence from school was due to seizures (16.9%), doctor visits (22.5%), difficulties in adaptation at school

(11.3%: noise intolerance, concentration problems, conflicts, etc.). Physical training was not attended by 23.9% of children, with no relationship to the severity of the disease. In 64.8% cases no impact on the leisure activities were reported. For those who reported restrictions in leisure activities, this was because of parental fears (36%) and the doctor's advice (24%). The majority of friends were aware of the patient's epilepsy in 76.2% of cases, and 23.8% of respondents told that nobody or just the best friend knew about it. Sufficient knowledge about epilepsy was claimed by 77.5% of children, and the rest admitted that they need to know more.

Conclusions. The majority of the children with active epilepsy have sufficient social adaptation and integration, epilepsy does not seem to have a great impact on their schooling, social communication and leisure activities. The restrictions, if any, are mostly due to fears from parents or medical professionals.

SUDEP: THE ATTITUDES OF ADULT AND CHILD NEUROLOGISTS

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Background. Recently, much attention is being given to the awareness of the possibility of sudden unexpected death (SUDEP) in patients with epilepsy.

Objective. To determine the attitudes of adult neurologists (AN) and child neurologists (CN) towards the provision of information about SUDEP to their patients with epilepsy and their families.

Methods. AN and CN were asked to fill out closed questionnaires containing questions about their personal clinical experience in dealing with patients with epilepsy in general, and in talking to patients/families about SUDEP, also about their opinion on why, when, how, to which patients, and by whom the information about the possibility of SUDEP should be provided.

Results. The questionnaires were answered by 107 specialists: 59 (55.1%) AN and 48 (44.9%) CN. The medium duration of clinical practice of AN was 19.8±11.3 years, and that of CN – 24.4±8.9 years. Most of their patients were informed about SUDEP only by 6 (10.2%) AN and 3 (6.2%) CN, individual patients were informed by 19 (32.2%) AN and 32 (66.7%) CN ($p<0.05$), never talked about this 34 (57.6%) AN and 13 (27.1%) CN ($p<0.05$). Thirty (50.8%) AN and 25 (52.0%) CN ($p>0.05$) admitted that the patients should be aware of the possibility of SUDEP, but more of them valued the importance of the awareness of family members: 42 (71.2%) AN and 41 (85.4%) CN, respectively ($p<0.05$). The question when this information should be provided was answered as follows: as soon as possible – 16 (27.1%) AN and 15 (31.2%) CN, only in cases of refractory epilepsy – 34 (57.6%) AN and 30 (62.5%) CN and only if the patients ask – 9 (15.3%) AN and 3 (6.3%) PN ($p<0.05$). The best way to give a information is “during the visit to the doctor” – 20 (33.9%) AN and 25 (52.1%) CN ($p<0.05$), “only in written form” – 12 (20.4%) AN, and none of CN; “both ways” – 27 (45.7%) AN and 23 (47.9%) CN.

Conclusions. Only a half of both adults and child neurologists believed that patients should be informed about the possibility of SUDEP. The majority of neurologists did not inform about SUDEP their patients/families or informed only individual patients. About a half of both adult and child neurologists thought they should inform about SUDEP only patients with refractory epilepsy, and nearly half of all specialists believed that patients should receive information on SUDEP in written form and discussed during the visits to the doctor.

CORRELATION OF ULTRASONOGRAPHY AND DOPPLER SONOGRAPHY PARAMETERS WITH DEVELOPMENTAL OUTCOME AT 1 YEAR OF AGE IN NEONATES WITH HYPOXIC-ISCHAEMIC BRAIN DAMAGE

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Background. Perinatal asphyxia is one of the most common causes of mortality and morbidity in full-term neonates. Early identification of hypoxic-ischemic (HI) brain damage could help to predict prognosis and to plan timely interventions.

Objective. To determine the correlation of ultrasonography (USG) and Doppler sonography (DSG) findings in neonates with HI brain damage with neuromotor and mental development outcome at 1-year of age.

Methods. The case group (n=78) consisted of neonates 37 weeks of gestation who experienced perinatal asphyxia or hypoxia. The control group (n=47) consisted of healthy neonates. USG and DSG were performed every day during the first five days of life. Cerebral blood flow parameters (peak systolic velocity (Vs), end-diastolic velocity (Vd) and resistance index (RI)) were measured in the anterior cerebral arteries (ACA) and the medial cerebral arteries (ACM). Neuromotor and mental development was assessed at the age of 12±1 months using the standardized neuromotor evaluation scale and Bayley Scale for Infant Development. The relationship of HI damage detected using USG and cerebral blood flow velocity parameters (Vs, Vd, RI) evaluated using DSG during first five days of life with neuromotor and mental development at 1 year of age was investigated.

Results. 83% (n=5) of subjects who were found having brain oedema, thalamus and/or basal ganglia, cerebellum and brainstem damage on USG died before the age of 1 year (p<0.001). Half (n=3) of subjects with oedema and thalamus and/or basal ganglia damage developed severe mental retardation and spastic quadriplegia (p<0.001). In subjects with spastic quadriplegia the means of Vd and RI values registered on ACA on the day 2 significantly differed from those with normal development: Vd was higher (>20 cm/s) (p<0.05) and RI lower (<0.55) (p=0.05). The mean Vd values in ACA registered during the first three days of life in subjects who developed severe mental retardation were significantly higher (>20 cm/s) (p<0.05) and mean RI values in ACA and ACM were significantly lower (<0.55) (p<0.05) than in subjects with normal development or with mild mental retardation.

Conclusion. Brain damage detected using USG and cerebral blood flow parameters (higher Vd and lower RI) evaluated using DSG during the first three days of life were associated with poor outcome at 1 year of age.

HOW SCHOOL-AGED CHILDREN EXPRESS THEIR HANDEDNESS

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Background. Evaluation of handedness belongs to the routine neurological examination. The idea of this study arose from the observation that children answer in various ways to

the question 'which is your main hand?'. The goal of this study was to evaluate the different ways used to express handedness and the evolution of gestural and verbal answers with age.

Method. Two hundred school children aged 6 to 18 years were asked the question 'which is your main hand?' during their neurological examination in outpatient clinic of Neurology Department of University Hospital of Lithuanian University of Health Sciences. Only children who attended a regular school and had no focal neurological symptoms were included in the study. Children were assigned to 3 groups according to their age: group I – 6 to 9.9 years, group II – 10 to 13.9 years, group III – 14 to 18 years. According to the response – gestural, verbal, or both, the answers were classified into 3 types: 'Shows', 'Tells', 'Shows and tells'. The latter type was further divided according to the sequence of the response: 'Simultaneously', 'Shows 1st', 'Tells 1st'. Rapidity of the 3 types of answers was assessed and classified as 'Immediate', 'Delayed' or 'Very delayed'. Eventually answers were analyzed according to gender and handedness.

Results. Study included 105 boys and 95 girls; 182 were right-handed, 18 left-handed. Group I included 44, group II 80, group III 76 subjects. The number of subjects according to the type of answer was: 46 'Shows', 59 'Tells', 95 'Shows and tells'. The percentage of these answer types varied significantly among age groups. The percentage of children who answered simultaneously increased with age. Concerning the rapidity to answer, 149 answers were 'Immediate', 30 'Delayed', 21 'Very delayed'. The proportion of children answering immediately increased with age. The highest percentage of children who responded with delays belonged to group I. Result parameters were not statistically different depending on gender or handedness.

Conclusion. The way school-aged children express their handedness varies with age. The sole gesture, which is the way used by most young children, is progressively replaced by a bimodal response in most. Purely verbal response concerns a minority of children irrespective of their age. Rapidity of expressing handedness increases with age. Gender and handedness do not influence the answers significantly.

ETHNICAL FEATURES OF NEURODEVELOPMENT OF INFANTS WITH FETAL ALCOHOL SYNDROME

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Fetal Alcohol Syndrome is a dramatic disease with social, regional and ethnic features (Y Senecky et al., 2011; PA May et al., 2006, 2011; DL Viljoen et al., 2005). As the population in many Russian regions is nonhomogenous, we can speak about regional and ethnic features of FAS manifestation. One of the bright examples is Republic of Sakha (Yakutia) – RS.

Aim of investigation – to study neurodevelopment of children with FAS during the first 7 years of life in different regions and ethnic groups in RS.

Materials and methods. Our study was carried out at 3 medical institutions and 12 uluses in RS. Observation by means of 4-digit Diagnostic Code, routine and age-dependent developmental neurological assessment, brain ultrasonography, MRI, Denver test (DDST, 1967).

FAS prevalence study (in view of ethnicity) in 12 uluses, 2 orphanages and National Pediatric Centre showed that FAS and FASD detection is more frequent among native-born babies (8.7:1000 and 4.8:1000 in uluses, 20 and 10% in orphanages accordingly).

We observed 58 children with FAS aged from 1 month to 7 years old, in 3 groups according to the ethnicity: I group – 26 Caucasians, mainly the Slavs; II group – 19 Sakha babies; III group – 13 natives: Evens, Evenks.

The study showed: Sakha children failed fine motor skills tests more frequently than the native ones. The native-born babies had better social adaptation figures than the Caucasians ($\chi^2=10.26-4.10$; $p=0.0014$). Weight loss, body length, head circumference were associated with neurological signs (hypodynamia, nystagmus, gait disturbance) ($r=0.33-0.44$; $p=0.004-0.03$), social maladjustment, speech disorders, gross and fine motor skills disorders, detraction ($r=0.27-0.52$; $p=0.007-0.04$). We found significant correlation between short palpebral fissure and mental retardation ($r=0.64$; $p=0.001$).

Conclusion. Our investigation shows the necessity of taking into account ethnical and regional features of FAS manifestation in neurodevelopmental study.

PREVALENCE AND CLINICAL PICTURE FETAL ALCOHOL SPECTRUM DISORDERS IN BABIES HOMES OF LITHUANIA

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Introduction. According to the WHO, Lithuania is in a 12th position of adult (15+) per capita alcohol consumption in the world. There is no data on the prevalence of fetal alcohol spectrum disorders (FASD) and concomitant disorders in the country.

Objectives. The purpose of this study was to determine the prevalence of FASD and concomitant somatic disorders in children living in Babies Homes in Lithuania.

Materials and methods. A cross-sectional study was performed in 5 Babies' homes (orphanages for children up to 3–4 years) where 337 children were examined. The paediatric examination included anthropometric measurements and phenotypic screening. Revised IOM criteria for the diagnosis of FASD were used.

In addition, 16 children diagnosed with fetal alcohol syndrome (FAS-16 group) underwent more detailed clinical examination: cranial CT scan, audiometry, auditory brainstem response (ABR), abdominal ultrasound, ophthalmoscopy, sleep EEG, evaluation by endocrinologist.

Results. FASD was identified in 40% (134) of children; FAS was found in 22%, partial FAS in 7%, and alcohol-related neurodevelopmental disorders in 11%. FASD diagnoses prior to the study have been suspected in 29.9% of these children.

In the FAS-16 group, the following findings have been recorded: cerebral dysmorphism in 7/16 (43.8%); hearing disorders confirmed by audiography in 9/14 (64.3%), with a medium or severe hearing disorder in five children; abnormal ABR in 7/13 (53.8%); retinal changes in 5/16 (31.2%); nonspecific changes on sleep EEG in 9/16 (56.3%); one child was diagnosed with hepatomegaly and one with hypopituitarism.

Conclusion. This study, being the first one in Lithuania focused on FASD, has shown that FASD is highly prevalent in children living in orphanages, however, underdiagnosed. Also, awareness and knowledge of specialists in the topic seems to be insufficient. Hearing disorder is worthy of note for its high prevalence in FASD population and for its possible impact on neurodevelopment.

BOTULINUM TOXIN A THERAPY FOR AMBULANT CHILD WITH CEREBRAL PALSY: IMPLEMENTING THE NEW APPROACH IN CHILDREN'S REHABILITATION HOSPITAL "LOPSELIS"

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Background. Repeated multilevel Botulinum toxin type A (BTX-A) treatments, started at an early age, combined with casting, orthotic management and physiotherapy could help to prevent muscle contractures and severe bony deformities and so to reduce and delay surgery until gait is matured in children with cerebral palsy (CP).

The effectiveness of BTX-A therapy has improved in the past 20 years, due to several reasons. 3-dimensional gait analysis as a tool for selection of most affected muscles and ultrasound as guidance for BTX-A therapy are two of them.

Objective. To present a case of BTX-A use in an ambulatory child with CP in order to present new diagnostic and treatment options that are implemented in Children's Rehabilitation Hospital 'Lopselis'.

Materials and methods. The 4-year-old girl with spastic diplegia, GMFCS – level 3, was evaluated by clinical examination and 3-dimensional computer-based gait analysis before and 6 weeks after a BTX-A injection.

The girl underwent BTX-A injections to gastrocnemius, soleus, semitendinosus, semimembranosus, gracilis and adductor longus muscles bilaterally. BTX-A injections were performed under ultrasound guidance.

Serial stretching casts were applied to both ankles joint for a period of three weeks. Hinged ankle-foot orthoses and outpatient individually designed physiotherapy plan were prescribed.

Results. Clinical examination and gait analysis 6 weeks after treatment demonstrated improvement in gait pattern and posture. Gait analysis data demonstrated that knee extension in preparation for initial contact and at initial contact, maximal dorsiflexion in stance and ankle range of movement during push off, excessive hip adduction in stance also has improved.

Conclusions. The usage of 3-dimensional gait analysis for proper muscle selection, an accurate injection technique, integrated treatment approach, when BTX-A is combining with the other conservative treatment options, ensures the success of BTX-A therapy.

USE OF PAIN-KILLERS AND CNS DRUGS FOR 8 TO 15 YEARS OLD CHILDREN WITH CEREBRAL PALSY IN DENMARK

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Background. Three out of 4 children with cerebral palsy (CP) are in pain; 1 out of 4 has active epilepsy; 1 out of 4 has a behavior disorder; 1 out of 5 has a sleep disorder. Use of oral medication for spasticity, pain, epilepsy, sleep or behavior disorders has not been investigated in a population sample before.

Aim. To describe the use of oral CNS drugs and pain-killers in every level of Gross Motor Function Classification System (GMFCS) in a population of 8 to 15 years old children with CP.

Method. Web-based parental questionnaire about oral CNS drugs and pain-killers was developed as a part of national survey on treatment methods of CP (cross-sectional design). Proportion of children using medication daily and not daily was calculated in every GMFCS level with 95% CI.

Results. Use of pain-killers increased from 6% in GMFCS level I to 62% in GMFCS level V, but only 1% of parents reported giving pain-killers daily. Use of spasmolytic drugs was reported from 4% to 74%, antiepileptic drugs from 10% to 60%, sleep inducing drugs from 1.3% to 31.7% in GMFCS levels I to V. Only 2% of parents reported use of CNS stimulants/ADHD medication and 0.5% reported use of antidepressants.

Conclusion. Parents reported very few children using pain-killers daily in contrast to a big proportion of CP children reporting daily pain in other studies. Use of CNS drugs was as expected from the clinical point of view.

COCHLEAR IMPLANTS IN CHILDREN WITH MENTAL RETARDATION

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Aim. To study results and benefits of cochlear implant (CI) in children with different degrees of mental retardation.

Materials and Methods. 24 children aged 4 to 14 with different degrees of mental retardation 12 to 24 months after CI participated in the study. The outcomes were assessed using questionnaires, parent satisfaction interviews. Listening abilities and ways of daily communication also were analysed.

Results. 17 out of 24 children used CI regularly, parents satisfaction ranged from 5–10 points (0 – min., 10 – max). The reactions to sound stimuli in the pure tone audiometry were from 15–40 dB. Psychological assessment showed that all children had mental retardation from mild to severe degree. 5 children out of 24 use spoken language in their daily communication, 10 – sign language, and 9 – simple signs and alternative communication.

Conclusions. The CI for mentally retarded children provided benefits regarding listening abilities, speech perception and ways of communication. Parent satisfaction was also remarkable.

INFLUENCE OF ENVIRONMENTAL FACTORS ON THE ONSET OF ANOREXIA NERVOSA

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The combination of psychological, biological and environmental factors leads to long-term dieting and weight loss which may contribute to the development of anorexia nervosa. Adolescence, followed by hormonal changes and feelings of stress, anxiety and low self-esteem, susceptibility to influence of the media, social factors may increase the risk of early onset of anorexia nervosa.

Aim. To evaluate environmental factors that might influence the early onset of anorexia nervosa.

Methods. Twenty four case histories were analyzed retrospectively. All patients were diagnosed with anorexia nervosa and treated in Psychiatry department of Hospital of Lithuanian University of Health Sciences Kauno klinikos between 2010–2012. Patients were divided into 2 groups according to the age of onset. Social environmental factors, leisure activities, physical exercise and addictions were evaluated. Data analysis was performed using ² test and Pearson's r coefficient, predetermined significance level $p < 0.05$.

Results. The sample consisted of 24 female patients (mean age 19 ± 9 yr), who were divided into 2 groups according to the age of onset: I – 18yr (n=; 79.2%); II – >18yr (n=5; 20.8%). Significant negative correlations were found between the age of onset and the following factors: the presence of siblings ($p=0.016$); parental employment status ($p=0.048$); leisure activities ($p=0.045$) and physical exercise ($p=0.02$). There was a positive correlation among psychoactive substance abuse in home environment and age of onset ($p=0.048$). Pre-school environment (including the environment the child grew up in, such as home, kindergarten and boarding schools) presence of physical abuse in family, parental expectations, marital status of parents, absence of close friends and academic performance did not differ between age groups.

Conclusions. 1. Presence of siblings, parental employment, engagement into leisure activities and physical exercise are related to early manifestation of anorexia nervosa. 2. Psychoactive substance abuse in home environment is not related to early onset of anorexia nervosa. 3. Pre-school environment, presence of physical abuse in family, parental expectations, marital status of parents, absence of close friends and academic performance did not increase the risk of early onset of anorexia nervosa.

CASE REPORT: ANOREXIA NERVOSA IN A 14-YEAR-OLD BOY

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The literature on primary anorexia nervosa (AN) in males mainly focuses on its rarity compared to female AN, resulting in incomplete clinical and empirical understanding of the disease and its risk factors. Despite a certain clinical similarity with female AN, it is reported to have some special traits. At one point male AN is thought to demonstrate more severe psychopathology. On the other hand males have been described to be more resistant to treatment.

Case Description. A 14yo boy was referred for treatment of disrupted eating behavior, rapid weight loss, intrusive thoughts and decreased mood. He has lost 40 kg (44.4%) of weight in 3 years. Patient was bullied at school for being overweight (BMI was 31) and his sexual orientation. He started to restrict the amount and type of food, excessive exercise, often self-weighing and checking his appearance in the mirror. The patient was consulted by a psychiatrist for 11 months, was prescribed medications, attended psychotherapy, but the treatment was ineffective. The patient's father had died when he was 1yo, mother re-married and went through a divorce. She has been on a diet for a year and lost 10 kg, which the patient supported. The patient demonstrated a capricious nature since childhood, demanding for things to be done his way and refusing to accept other outcomes. The patient attended dance classes which he quit after receiving criticism. Then he started to spend leisure watching TV and reading magazines that focus on clothing and fashion. Physical exam-

ination revealed bradycardia (HR 49), low BP (110/65 mmHg), hypothermia and hypothyreosis. Blood tests revealed anemia, hyperkalemia. During the 20-day hospital stay the boy collaborated actively, improved his eating pattern, by the end managing to finish the whole meal, gained 4 kg. Cognitive behavioral, individual and family psychotherapies were applied. The mother remained uncritical during family therapy and refused treatment on the patient's behalf.

Discussion. This case represents typical male AN with a variety of risk factors: early onset of the disease, living in an environment fixated on physical appearance, media featuring 'perfect' people, being overweight in childhood and a vegetarian way of life, which is followed by a severe pathology and is resistant to treatment. Following the increasing numbers of male anorexia, attention to the listed risk factors in adolescent boys should be given, and a psychiatric consultation sought if AN is suspected.

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