and 3.39% were found to have a perinatal/antenatal brain injury etiology. Of the 906 patients who had records for the seizure history, 39% were diagnosed with epilepsy and 93.77% of these patients had clinical seizures and an electroencephalography-determined seizure. The rate of global growth retardation was determined as 88.6% in the patients with developmental evaluation abnormal by DENVER II developmental screening test. When DENVER II findings of the patients with growth retardation were evaluated in terms of area, it was found that 91.1% of the patients had language, 82.7% had personal and social, 89.3% had gross motor and 87.1% had thin motor field regression. The patients with WISC-R were found to have a mild intellectual disability rate of 51.1% (n = 24), a moderate mental disability rate of 44.7% (n = 21) and a severe mental deficiency rate of 4.3% (n = 2) determined. 29.72% of patients who underwent cranial MRI showed abnormal findings. Cerebral anomaly was found in 94.3% of patients with abnormal cranial MRI findings, 16.7% had brain stem dysplasia, 11.6% had cerebellar anomalies, 1.4% had pituitary gland hypoplasia and 0.2% had meningoencephalocele were detected. Therefore, microcephalic patients should be closely monitored in terms of epilepsy, mental insufficiency and global growth retardation, and counseling should be provided to their families. In addition, it was found that cranial MRI was important for the clarification of some etiologies.

P495 IS NARCOLEPSY INCIDENCE INCREASING OR SYMPTOMS JUST BETTER RECOGNISED?

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Background Narcolepsy is a rare and under-recognised disease characterised by excessive daytime sleepiness, disturbed nocturnal sleep, hallucinations, and cataplexy, with a major impact on quality of life. Diagnosis can be challenging. We present a series of recent children presenting to a district general hospital over a 2.5 year period with chronic symptoms ultimately diagnosed with narcolepsy.

Patients *Patient 1:* Female(12y) GP referral with 2-year history of increased sleepiness and falling asleep during meals. Later developed cataplexy-'when I laugh, I go weak suddenly'. Swine-flu vaccine 6 years previously.

Patient 2: Female(8y) GP referral with 18-month history of excessive day-time sleepiness and vivid nightmares. Narcolepsy was considered when seen in clinic initially but significant academic decline subsequently prompted formal diagnosis (9 months after referral). Laugh induced catatonia (eyes roll, tongue protrusion and lower limb weakness). Pandemrix H1N1 vaccine 3 years previously. Sleep study noted abnormal sleep latency and sleep onset REM periods. Improvement with methylphenidate.

Patient 3: Male(10y) emergency department (ED) presentation with a 2-month history of excessive day time sleepiness and increased appetite. Falling asleep while standing occasionally. No cataplexy. Sleep study abnormal (2.3 minutes sleep

latency and SOREM periods in 3/4 naps). Treatment with sodium oxybate.

Patient 4: Female(5y) ED presentation with 2-month history of daily episodes of eye rolling and sudden collapse, weeks following MMR. Initial diagnosis and treatment for epilepsy. Video review reveals clinical diagnosis of cataplexy; focused history revealed excessive daytime sleepiness. Treatment with stimulants (methylphenidate) improved sleep symptoms; considering sodium-oxybate for cataplexy.

Patient 5: Male(15y) GP referral with 3 year history of excessive day-time sleepiness, struggling to keep up academically. Upon laughing his mouth droops open with tongue protrusion (cataplexy). No swine-flu vaccine. Treatment pending investigations.

Patients 1-4 had undetectable CSF hypocretin levels (except patient 2; failed LP) and positivity for the HLA-DQB1*0602 association in keeping with narcolepsy. Patient 5 is awaiting specific investigations. Sleep studies are pending for 3 patients. Secondary causes of narcolepsy were out-ruled.

Discussion Delay in narcolepsy diagnosis is associated with worse outcomes. While narcolepsy is thought to be under recognised, these 5 patients were suspected upon referral to paediatrics and management instigated quickly; however focused history and increased awareness is required. Our series and review of a national narcolepsy clinic suggests increased incidence and early detection in children. A national narcolepsy service (currently Temple-St-Hospital, Dublin) facilitates management, as symptoms and co-morbidities can be debilitating and life-long. Vaccine association remains controversial.

P496 X- LINKED INFANTILE SPINAL MUSCULAR ATROPHY (SMAX2) CAUSED BY NOVEL C.1681G>A SUBSTITUTION IN THE UBA1 GENE, EXPANDING THE PHENOTYPE

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Aims We report the case of a male infant who presented following a normal pregnancy with typical symptoms of X-linked infantile spinal muscular atrophy including hypotonia, weakness, areflexia and respiratory insufficiency, however contractures were absent. X-linked infantile spinal muscular atrophy (SMAX2), OMIM 301830, is a rare, severe form of spinal muscular atrophy, caused by variants in the Ubiquitin like modifier-activating enzyme 1 (UBA1) gene1. Clinical features reported to date include marked hypotonia, areflexia, arthrogryposis, contractures, myopathic facies, tongue fibrillations and cryptorchidism2. To our knowledge, this is the first reported case of SMAX2 to present without contractures.

Methods This infant was born at 40+4 weeks following a normal pregnancy to non-consanguineous Irish parents. Intermittent stridor was noted following delivery, laryngomalacia was diagnosed following review by ENT. At 8 weeks old there was an acute event at home with colour change and poor feeding for 2 days. On presentation to ED, there was marked increased work of breathing, weakness, absent deep tendon reflexes, hypotonia, myopathic facies and weak cry. Eyes were bright and there was no ophthalmoplegia or contractures. A significant family history of neuromuscular disease on the patient's maternal side was noted, with several male relatives all dying before the age of six months. Creatine Kinase was

mildly elevated, MRI Brain was normal and neurophysiological testing revealed a diffuse motor neuronopathy. Genetic testing for SMN1 gene was normal. UBA1 sequencing revealed a maternally inherited hemizygous familial variant [c.1681G>A p. (Asp561Asn)], which has not been previously reported.

Results To our knowledge, only six families with SMA2 have been confirmed at a molecular level to date.

Conclusions After ruling out 5q SMA, X-linked infantile spinal muscular atrophy should be considered in male patients with anterior horn cell disease in the absence of contractures.

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REHABILITATION OF CHILDREN WITH SPASTIC AND DYSKINETIC FORMS OF CEREBRAL PALSY THROUGH TRANSCRANIAL EXPOSURE AND BIOLOGICALLY FEEDBACK

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Movement disorders are determining clinical syndrome of cerebral palsy (CP). The prevalence of CP in Russia is 2.2–3.3 cases per 1000 newborns; 85–90% of them are spastic forms. Complex treatment of children with spastic CP includes Botulinum toxin type A as the main component - it reduces spasticity, which optimizes the subsequent rehabilitation. Combination of botulinum therapy with physiotherapy increases the effectiveness of management CP in children. Transcranial magnetic stimulation was carried out on the 'Amo Atos' device using the add-on device 'OGOLOVYE', according to our own method with a bitemporal arrangement of emitters, with increasing frequency in subsequent procedures from 5 Hz to 10 Hz, for 10 minutes. Transcranial magnetic stimulation was combined with physical exercise on the device with biofeedback 'MOTomed gracile' according to an individual scheme after botulinotherapy.

Aim To evaluate the effectiveness of combined use of transcranial magnetic stimulation and physical exercise using the biofeedback device with simultaneous neurometabolic medical therapy, exercise therapy, massage after administration of botulinum toxin type A.

Materials and methods The case-control study included 30 patients 2–14 years old with spastic and dyskinetic CP, comparable by sex, age, degree of motor disorder. The degree of motor disorders was determined by Gross Motor Function Classification System. Evaluation of effectiveness – according to muscle strength scale; tone was estimated by Modified Ashworth Scale; detailed assessment of motor skills according to GMFM-66/88 table, considering the quantitative indicator of motor development. Patients of both groups received botulinum therapy, nicotinic acid, massage, exercise therapy. The main group received physical load on the device 'MOTomed gracile', transcranial magnetic stimulation; the control group underwent sinusoidal magnetic fields.

Results A 1 point decrease in muscle tone by 10 days was significantly achieved in 92% of children of the main group and in 75% of the children in the control group; change in

strength was found in 25% of children in the main and 12% of the control group; improvement in motor skills was observed in all patients, in the main group by 5.5%, in the control group - by 3%.

Conclusions The study shows high efficacy of botulinum toxin, a good antispastic effect in both groups. Improvement of motor skills and new motor patterns occurs in a fairly short time only with an active rehabilitation program. It confirms the effectiveness of transcranial magnetic therapy and physical activity on the device with biofeedback 'MOTomed gracile'.

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BOTULINUM TOXIN THERAPY IN PATIENTS WITH CEREBRAL PALSY UNDER TWO YEARS

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Background and aims The spasticity is a major clinical sign of cerebral palsy (CP). The most effective and safe method of local spasticity decrease is the botulinum toxin A (BTA) therapy (evidence level A). Studying of early BTA injections in complex rehabilitation of children with CP is relevant due to the detection of wide range of secondary orthopedic problems in patients with early and severe spasticity.

The objective was to determine the most common spasticity patterns for botulinum toxin therapy in patients with cerebral palsy under 2 years and to calculate the BoNT-A mean effective dose for the first injection.

Patients and methods 60 patients (38 boys, 22 girls) aged from 15 up to 23 months (median age 21 months) with spastic forms of CP. Among all patients 20 had tetraparesis, 30 – diplegia, 10 – hemiparesis. The prevalence and intensity of muscle hypertonia was estimated in every limb functional segment with the Modified Ashworth scale (MAS). Injections were performed 'off-label' after the written consent of parents or representatives and with the agreement of the local independent ethics committee. Ultrasound control and Onabotulinum Toxin A were used in this study.

Results Spasticity patterns were divided as shown below: forearm pronation in 51 patients (85%), equinus foot deformity – 47 (78,3%), hip adduction and internal rotation – 41 (68,3%), elbow flexion – 39 (65%), hip adduction – 33 (55%), pollex adduction – 21 (35%), rectus syndrome – 11 (18,3%), shoulder adduction and internal rotation – 10 (16,7%), elbow extension – 8 (13,3%), in 5% and less were knee flexion, wrist flexion, fingers flexion. Regardless of early age 57 (95%) patients required multi-level injections. The total dosage for all patients ranged 50–230 U (4,8–23 U/kg). In each injected muscle tone reduced for at least 1 MAS point. Ranges of Onabotulinum Toxin A dosage per muscle are presented in the Poster.

Discussion Definition of the most essential spasticity patterns and their influence on the child movement development is among the key points for the target muscle selection during BTA therapy. Presented dosage ranges allowed us to reach significant decrease of spasticity in all patients.