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## Interventions and Management

### 1. Dev Neurorehabil. 2013 Jan 16. [Epub ahead of print]

#### Reliability of retrospective assignment of gross motor function classification system scores.

Mayson TA, Ward V, Davies KR, Maurer J, Alvarez C, Beauchamp R, Black A.

Shriners Gait Lab, Sunny Hill Health Centre for Children , Vancouver, British Columbia , Canada.

Objectives: To assess "alternate forms" reliability and inter-rater reliability of Gross Motor Function Classification System (GMFCS) scores. Methods: Fifty randomly selected children with cerebral palsy were divided into two groups: (1) GMFCS score assigned during gait assessment ("GMFCS previously assigned") and (2) no GMFCS score assigned. Using database information, two physiotherapists independently determined GMFCS scores for 25 children from the "previously assigned" group, and 25 from the "no score assigned" group. Therapists compared their recently assigned scores for the "previously assigned" group, discussing discrepancies until attaining agreement. This group's consensus scores were compared to GMFCS scores assigned at time of actual assessment to calculate "alternate forms" reliability. Results: Between-therapist agreements were kappa = 0.84 for "GMFCS previously assigned" group and 0.95 for "no GMFCS assigned" group. Kappa agreement between direct assessment and retrospectively assigned scores for the "GMFCS previously assigned" group was 0.79. Conclusions: Retrospective GMFCS scores can be reliably assigned.

[PMID: 23323825](#) [PubMed - as supplied by publisher]

### 2. Handb Clin Neurol. 2013;110:443-59. doi: 10.1016/B978-0-444-52901-5.00038-1.

#### Cerebral palsy.

Kent RM.

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Cerebral palsy affects movement and posture causing activity limitation; it is a lifelong condition, with foreseeable complications. There are evidence-based interventions that will prevent participation restriction. Childhood interventions are generally delivered within multidisciplinary rehabilitation programs. Sadly young adults are often not transferred to an appropriate multidisciplinary adult neurodisability service. An unexplained neurological deterioration should warrant further investigation. Pain is an important underreported symptom and

musculoskeletal complaints are prevalent. Disabled adults have less participation socially, in employment, marriage, and independent living related to health problems, discrimination, or lack of access to information, support, and equipment. Evidence-based interventions include a variety of modalities at all International Classification of Functioning, Disability, and Health levels to include support and adaptations. Rehabilitation interventions that have been shown to be effective include surgery in childhood, ankle-foot orthoses, strength training, and electrical stimulation. Management of spasticity is beneficial and has an evidence base. Orthotics and casting are also used. Systematic reviews of upper limb therapies also show the benefit of physical therapy exercise, strengthening, fitness training, and constraint therapy. Occupational therapy has a weaker evidence base than in other disabling conditions but many modalities are transferable. Speech therapy is effective although no specific intervention is better. Psychological wellbeing interventions, including improving self-efficacy, health knowledge, and coping skills, are beneficial. Management of continence, nutrition, and fatigue promote wellbeing.

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[PMID: 23312663](#) [PubMed - in process]

**3. Ann Phys Rehabil Med. 2012 Dec 29. pii: S1877-0657(12)01306-1. doi: 10.1016/j.rehab.2012.11.004. [Epub ahead of print]**

**Sagittal X-ray parameters in walking or ambulating children with cerebral palsy.**

Deceuninck J, Bernard JC, Combey A, Leroy-Coudeville S, Morel E, Loustalet E, Chaleat-Valayer E, Berthonnaud E.

Croix-Rouge française, CMCR des Massues, 92, rue du Dr-Edmond-Locard, 69005 Lyon, France; Laboratoire de physiologie de l'exercice, université de Lyon, 42000 Saint-Étienne, France. Electronic address: kine.mpr.enfants@cmcr-massues.com.

Adolescents with cerebral palsy (CP) who walk or ambulate often have an abnormal clinical and radiological spinal profile during pubertal growth compared with adolescents of the same age without neuromotor impairments. Therefore, in the following study, we aimed to conduct a radiological assessment of static data on the lumbar-pelvic-femoral complex in ambulatory children with CP to compare these data with those of an asymptomatic population. The CP population was comprised of 119 children and the asymptomatic population was comprised of 652 children. The large format (30×90cm) sagittal X-rays were taken while subjects were in a comfortable position in which knees and hips were in maximal extension. Analyses were performed using Optispine(®) software to measure the parameters of an X-ray of the profile of the spine, pelvis and femurs. Comparing, the two populations, we found no difference in the shape parameter (pelvic incidence) but we did find significant differences in the positional parameters (pelvic tilt and sacral slope) of the pelvis. We found a difference in the curvature and orientation of lumbar lordosis as well as in the number of vertebrae involved in the kyphosis and its orientation. There was also a significant difference in the C7 plumb line. We can say that the CP population is not structurally different from the control population, but that parameters become disturbed during growth. These disturbances should be identified and monitored so that changes can be detected early and progression can be prevented.

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**4. Am J Intellect Dev Disabil. 2012 Nov;117(6):442-54. doi: 10.1352/1944-7558-117.6.442.**

**Prevalence of falls and risk factors in adults with intellectual disability.**

Hsieh K, Rimmer J, Heller T.

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The purpose of this study was to examine the prevalence of falls and risk factors for falls in 1,515 adults (≥ 18 years) with intellectual disability using baseline data from the Longitudinal Health and Intellectual Disability Study. Nearly 25% of adults from the study were reported to have had one or more falls in the past 12 months. The

prevalence of falls increased with advancing age. A series of univariate and multivariate logistic regressions were performed to identify risk factors for falls in the full sample and in subsamples. The risk factors for falls in adults with intellectual disability are being female, having arthritis, having a seizure disorder, taking more than 4 medications, using walking aids, and having difficulty lifting/carrying greater than 10 lb.

[PMID: 23167484](#) [PubMed - indexed for MEDLINE]

**5. PM R. 2013 Jan 9. pii: S1934-1482(12)01712-1. doi: 10.1016/j.pmrj.2012.11.005. [Epub ahead of print]**

**Tolerability and Effectiveness of a Neuroprosthesis for the Treatment of Footdrop in Pediatric Patients With Hemiparetic Cerebral Palsy.**

Meilahn JR.

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**OBJECTIVE:** To assess the tolerability and efficacy of a commercially available footdrop neuroprosthesis for treatment of footdrop in children with hemiparetic cerebral palsy. **DESIGN:** A prospective, observational pilot study. **SETTING:** Marshfield Clinic, Department of Physical Medicine. **PARTICIPANTS:** Ten children, ages 7-12 years, with hemiparetic cerebral palsy, who use an ankle foot orthosis (AFO) for correction of footdrop. **METHODS:** Children replaced their AFO with a transcutaneous peroneal (fibular) nerve stimulation neuroprosthesis for 3 months. **MAIN OUTCOME MEASUREMENTS:** The ability to tolerate fitting and programming of the device, device-recorded wear time, a daily-use diary, satisfaction survey, and secondary measures, including passive range of motion and gait laboratory measurement of gait velocity and ankle kinematics. **RESULTS:** All 10 participants (100%) tolerated fitting and programming of the neuroprosthesis and wore the device for 6 weeks. Seven of 10 (70%) wore the device for the entire 3-month study period; 6 of 10 (60%) continued to use the device after study completion. Wear time varied from 2 to 11 hours per day. Tolerability and satisfaction were high; although 6 participants complained of "size" and "bulkiness" of the device, and 2 reported skin irritation. Gait velocity increased in 5 subjects (50%). Seven participants (70%) preferred the neuroprosthesis to their AFO. **CONCLUSION:** Analysis of the preliminary evidence suggests that electrical stimulation by a footdrop neuroprosthesis is tolerated well by children and is effective for the treatment of footdrop in children with hemiparetic cerebral palsy. Commercially available neuroprostheses may offer a promising alternative treatment option for children with footdrop.

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[PMID: 23313040](#) [PubMed - as supplied by publisher]

**6. Ann Phys Rehabil Med. 2012 Dec 14. pii: S1877-0657(12)01305-X. doi: 10.1016/j.rehab.2012.11.003. [Epub ahead of print]**

**A survey of medical and paramedical involvement in children with cerebral palsy in Brittany: Preliminary results.**

Sacaze E, Garlantezec R, Rémy-Néris O, Peudener S, Rauscent H, le Tallec H, Bernier-Francois V, Pichancourt D, Brochard S; et le Groupe de Recherche sur la paralysie cérébrale en Bretagne.

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**AIM:** To describe the amount of medical and paramedical involvement in a sample of Breton children with cerebral palsy as a function of the Gross Motor Function Classification System (GMFCS). **MATERIALS AND METHODS:** This is a transversal descriptive study. All children with cerebral palsy in Brittany were eligible. Parents who accepted to participate were asked to fill in a questionnaire regarding medical and paramedical involvement with their child. **RESULTS:** One hundred and thirty-three parents participated. 40.6% of the children were level I on the GMFCS, 20.3% II, 12.03% III, 13.53% IV and 13.53% were level V. Thirty-nine percent of the children took at least one medication (of which 43% were antiepileptic drugs). 33.1% of the children had received at least one injection of

botulinum toxin within the year. Forty-four percent used a mobility aid. Eighty-five percent of the children had at least one orthotic device, most often a night ankle-foot orthosis. The median number of rehabilitation sessions per week was 3.85 [0.5-11.5]. The frequency and type of sessions were mostly related to the GMFCS level.  
**CONCLUSION:** This study reports high levels of medical and paramedical involvement. Studies must attempt to define optimal practice.

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## **7. Handb Clin Neurol. 2013;110:229-37. doi: 10.1016/B978-0-444-52901-5.00019-8.**

### **Neurological rehabilitation: sexuality and reproductive health.**

Aisen ML.

Rancho Los Amigos National Rehabilitation Center, Downey, CA and Cerebral Palsy International Research Foundation, Washington, DC, USA. Electronic address: [maisen@ucpresearch.org](mailto:maisen@ucpresearch.org).

Sexuality is the embodiment of sexual and reproductive activities involving complex interactions among biological, psychological, and social systems. An individual's perception of their sexuality, as well as society's perception, can have an inestimable impact on self-esteem, and hence willingness to openly address these issues Earle S (2001). Disability, facilitated sex and the role of the nurse. *J Adv Nurs* 3: 433-440. Such barriers to communication represent a real challenge to practicing clinicians. However, advances in treatment options obligate the clinician providing care to those with neurogenic sexual/reproductive dysfunction to learn to communicate effectively about these issues, provide effective therapies, and refer patients to appropriate specialists. This chapter will address counseling, an overview of male and female sexual and reproductive physiological responses in the case of an intact nervous system, and a description of the impact of disorders of the nervous system on sexual function and reproductive health. Treatment options are also reviewed.

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[PMID: 23312644](#) [PubMed - in process]

## **8. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):24-8.**

### **Spasticity in children cerebral palsy: diagnosis and treatment strategies [Article in Russian]**

[No authors listed]

Spasticity in children cerebral palsy has its own peculiarities due to the presence of pathological tonic reflexes, pathological sinkinetic activity during arbitrary movements, disturbance of coordinative interactions of muscle synergists and antagonists, increase of total reflex excitability. Physiotherapeutic methods, massage, therapeutic exercises, kinesitherapy, biological feedback training (BFT), methods of orthopedic correction, neurosurgery are widely used in the treatment of spasticity. The use of botulinum toxin type A is a new effective approach to the treatment of spasticity that improves motor functions and quality of life of children with children cerebral palsy. It is being used in the treatment of children and adolescence in a polyclinic unit of the Moscow psychoneurological hospital since 2001. The experience of treatment with botulinum and wide implementation of this method indicated that botulinum toxin injections in the complex treatment of spasticity allow to optimize approaches to treatment of children and adolescence with children cerebral palsy and to increase significantly the quality of medical-social rehabilitation of patients.

[PMID: 23330188](#) [PubMed - in process]

**9. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):71-6.****Features of epileptiform activity on EEG in children with periventricular leukomalacia and cerebral palsy without epilepsy [Article in Russian]**

[No authors listed]

We have analyzed morphologic and chronologic characteristics of epileptiform activity, with account for repeated EEG-study during the follow-up, in patients with periventricular leukomalacia and children cerebral palsy without epilepsy. The high frequency of "benign epileptiform patterns of childhood" (BEPC) was noted. The epileptiform activity recorded by chronologic criteria corresponded to BEPC in 67% of children. The high probability of epileptiform activity of symptomatic character was identified in 33% of children. The results obtained in this study of the parameters of epileptiform activity could be of great importance for predicting the risk for the development of epilepsy and tactics of rehabilitation of motor disorders.

[PMID: 23330196](#) [PubMed - in process]**10. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):64-70.****Children cerebral palsy and epilepsy: approaches to treatment and rehabilitation [Article in Russian]**

[No authors listed]

Epilepsy is one of the most frequent and difficult for treatment co-morbid disease of cerebral palsy. In therapeutic aspect, the difficulty of the problem is defined by the necessity to combine the active restoration of motor disorders with a regime of antiepileptic treatment. It leads frequently to stopping the restoration process and aggravation of patient's motor disability. The diagnosis of epilepsy in the child with cerebral palsy should in no way discontinue the rehabilitation measures, albeit in case of the concomitant pathology a plan of rehabilitation scheme should be adjusted. The pharmacological control of epileptic seizures should be the first step of the new rehabilitation scheme. Epileptologists usually conduct the selection of multi-component antiepileptic treatment in patients with drug resistant epilepsy, however a neurologist of an outpatient clinics who follows up the patient in different stages of development and rehabilitation should play a key role. The authors suggest the general treatment tactics for children with cerebral palsy and epilepsy by the neurologist of the polyclinics.

[PMID: 23330195](#) [PubMed - in process]**11. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):56-63.****Family's adherence to treatment of the child with a neurological pathology [Article in Russian]**

[No authors listed]

Neurological diseases, mental disorders and inherited developmental abnormalities held a prominent place in the structure of primary children disability. Children cerebral palsy is the main cause of children disability, its prevalence reaching 2-3.5 cases per 1000 children. This disease has the noticeable negative consequences (both clinical and economic). Adherence to treatment means patient's compliance with doctor's orders including taking medicines as prescribed, sticking to diet and changing life style habits. While the adult patient plays an active role in doctor-patient alliance, the child patient interacts with the doctor together with his/her family. The authors consider the effect of different factors on the adherence basing on the analysis of 270 questionnaires completed by persons involved in the treatment of minor patients.

[PMID: 23330194](#) [PubMed - in process]

**12. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):52-5.****Logopedic and psychological-pedagogic maintenance of infants with cerebral palsy [Article in Russian]**

[No authors listed]

The problem of perinatal pathology of the nervous system in infants is getting more and more urgent due to the progressive growth of the frequency of cerebral disorders in newborns. This pathology leads to the development of severe motor and speech disorders which inhibit the development of communication skills and social adaptation of children in future. Perinatal factors cause not only neurologic and mental disorders but impede and distort the formation of pre-speech functions, basic for speech development, in infants thus worsening their disability. Patients with cerebral palsy make up 57% of children acknowledged as disabled. Therefore, logopedic diagnostic methods are needed to allow identification of the structure of pre-speech defects in infant period of life, prediction of the level of nervous-mental development, planning and realization of a correction-educational route from the first months of life.

[PMID: 23330193](#) [PubMed - in process]**13. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):47-51.****One of the approaches to psychological-pedagogical help to children with severe movement disorders [Article in Russian]**

[No authors listed]

The objective of the study was to work out an effective model of complex help to children with severe movement disorders. We examined 440 preschoolers with children cerebral palsy with severe movement disorders and 70 children with mild movement disorders. Functions of motor, emotional-personal and cognitive spheres and independence status with determination of 5 levels were studied in 47 patients. Three groups (from the group without concomitant (sensor, intellectual etc) disorders to the group with most severe disorders) were singled out. The authors characterize the model as an open integral system of methods, tools and ways providing the adaptation of children in response to external circumstances and changes in the state of patients. The creation of a correction-developing environment, consisting of 3 components: spatial-objective, technological (methodological) and social, is discussed. We present results of the development of children, evaluated by the following indices: general technique, sensory perceptive development, social adaptation, anxiety, cognitive activity, from 1997 to 2008. The 15 year follow-up demonstrated the stability of achieved positive results.

[PMID: 23330192](#) [PubMed - in process]**14. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):41-6.****Anesthesia in surgical treatment of patients with cerebral palsy (a review) [Article in Russian]**

[No authors listed]

Anesthesia in surgical treatment and anesthetization in post operative period in patients with children cerebral palsy (CCP) is considered as an actual problem due to some specific features. The authors review the current state of the problem in aspects of efficacy and setting priorities in the development of methods of anesthesia in patients with CCP. Some methods of anesthesia, positive and negative sides of their application in CCP are characterized.

[PMID: 23330191](#) [PubMed - in process]

**15. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):34-40.****Neurosurgery of the spasticity syndrome in children cerebral palsy [Article in Russian]**

[No authors listed]

The review is devoted to main neurosurgical approaches to the treatment of the spasticity syndrome in children cerebral palsy. Neurosurgical procedures are divided into destructive and neuromodulating. The former included posterior selective rhizotomy, selective neurotomy and destructive operations on subcortical brain structures. The latter group included electrostimulation of brain and spinal cord structures and implantation of pumps for the chronic intrathecal baclofen (Iioresal) infusion. Each method is considered in a historical aspect. Details of clinical application, positive and negative sides of the methods are described.

[PMID: 23330190](#) [PubMed - in process]**16. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):29-33.****Efficacy of botulinum toxin in the treatment of dynamic equinus and equinovarus foot deformities in children with hemiplegic cerebral palsy [Article in Russian]**

The objective of the study was to assess factors modulating the efficacy of botulinum toxin injections in the correction of dynamic equinus and equinovarus foot deformities in children with hemiplegic cerebral palsy. The efficacy of treatment was evaluated in 40 children. Clinical data including spasticity assessment by the original Ashworth scale, postural and gate changes were collected. Spasticity grade 4-5 by the Ashworth scale and retraction 90-120 degrees reduce the likelihood of successful correction of equinus contracture. Botulinum injections can be used as basic therapy. In cases of transient contracture more than 120 degrees, combined methods of treatment are recommended. The hindfoot varus less 30 degrees, which can be passively corrected before treatment and in the presence of the positive Coleman block test, more likely needs injection in triceps muscle for correction. If the hindfoot varus is more than 30 degrees and the Coleman block test is negative, the combined treatment is necessary.

[PMID: 23330189](#) [PubMed - in process]**17. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):18-23.****The effect of the reflex-load device Gravistat/Graviton on walk stereotype in patients with spastic diplegia [Article in Russian]**

[No authors listed]

We examined 6 children with spastic diplegia who were able to walk independently. We studied effects of different ways of the adjustment of the reflex-load device (RLL) Gravistat/Graviton according to spatial-temporal characteristics of lower extremity muscle work during walking (innervation stereotype) in children with cerebral palsy in the form of spastic diplegia. The positive effect of axial load of RLL on the innervation type of walking was demonstrated. The choice of a specific way of RLL adjustment should be based, in some cases, on the analysis of EMG dynamics.

[PMID: 23330187](#) [PubMed - in process]**18. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):14-7.****Changes of motor function in patients with cerebral palsy during the treatment using the intensiv neurophysiological rehabilitation system [Article in Russian]**

[No authors listed]

Changes in gross motor function during the intensive neurophysiological rehabilitation were studied in 61 patients,

aged from 2 to 15 years, with spastic forms of cerebral palsy. All patients were examined before and at the end of a two-week course of treatment using the Gross Motor Function Measurement GMFM-66 Item Sets test to calculate motor development scores. Statistical analysis indicates a significant increase in the level of motor development of children after treatment from 45.1 to 47.6 ( $p > 0.01$ ). The most significant progress was noted in patients at level II of Gross Motor Function Classification System. The score of motor development has increased from 66.2 to 69.6 ( $p < 0.01$ ). The results suggest the effectiveness of the Intensive Neurophysiological Rehabilitation System for the improvement of gross motor functions in patients with cerebral palsy. It is necessary to continue this study according to the requirements of evidence-based medicine.

[PMID: 23330186](#) [PubMed - in process]

#### 19. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):9-13.

##### The problem of rehabilitation of children cerebral palsy [Article in Russian]

[No authors listed]

The paper is devoted to the problem of early diagnosis and correction of development of children with perinatal lesion of the nervous system. Early rehabilitation treatment in children cerebral palsy (CCP) is necessary due to the plasticity of the child's brain and its universal ability to compensate for disturbed functions as well as due to the fact that the first two years of life are optimal for maturation of the speech functional system. Evidence for the differentiation of early, chronic and residual studies of CCP is presented. The authors emphasize the role of the pathology of the functional antigravitation system in the formation of motor-reflex disorders. Possibilities of pathogenetic treatment in each stage of disease are considered.

[PMID: 23330185](#) [PubMed - in process]

## Prevention and Cure

#### 20. BJOG. 2013 Jan 15. doi: 10.1111/1471-0528.12133. [Epub ahead of print]

##### Outcome of severe placental insufficiency with abnormal umbilical artery Doppler prior to fetal viability.

Simonazzi G, Curti A, Cattani L, Rizzo N, Pilu G.

Department of Obstetrics and Gynecology, St. Orsola Malpighi Hospital, University of Bologna, Bologna, Italy.

**OBJECTIVE:** To evaluate the outcome of pregnancies complicated by placental insufficiency and abnormal umbilical artery Doppler prior to viability. **DESIGN:** A retrospective cohort study. **SETTING:** Italy. **POPULATION:** Singleton pregnancies with fetal growth restriction and absence of end-diastolic velocities (AEDVs) in the umbilical arteries prior to 24 weeks. **METHODS:** A retrospective cohort study of singleton pregnancies with fetal growth restriction and AEDVs in the umbilical arteries prior to 24 weeks. **MAIN OUTCOME MEASURES:** Fetal growth restriction and AEDVs in the umbilical arteries prior to 24 weeks. **RESULTS:** Of 16 fetuses first seen at 20-23 weeks, only 12 survived and one of these developed cerebral palsy. Severe hypertensive disorders occurred in three mothers. In four women, the Doppler waveforms progressively improved and developed a normal pulsatility. These fetuses had a better outcome than those that had persistent alterations: they were delivered later (34 versus 28 weeks), had a larger birthweight (1598 versus 630 g) and developed fewer complications. **CONCLUSIONS:** Placental insufficiency with AEDV in the umbilical arteries prior to fetal viability is associated with a high probability of perinatal death and neonatal complications. However, progressive amelioration of Doppler indices occurs in a subset of women, and these fetuses have a much better outcome.

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**21. Brain. 2012 Oct;135(Pt 10):2980-93. doi: 10.1093/brain/aws240.**

**Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy.**

Klebe S, Depienne C, Gerber S, Challe G, Anheim M, Charles P, Fedirko E, Lejeune E, Cottineau J, Brusco A, Dollfus H, Chinnery PF, Mancini C, Ferrer X, Sole G, Destée A, Mayer JM, Fontaine B, de Seze J, Clanet M, Ollagnon E, Busson P, Cazeneuve C, Stevanin G, Kaplan J, Rozet JM, Brice A, Durr A.

INSERM, UMR\_S975 - CRICM, F-75013 Paris, France.

Mutations in the spastic paraplegia 7 (SPG7) gene encoding paraplegin are responsible for autosomal recessive hereditary spasticity. We screened 135 unrelated index cases, selected in five different settings: SPG7-positive patients detected during SPG31 analysis using SPG31/SPG7 multiplex ligation-dependent probe amplification (n = 7); previously reported ambiguous SPG7 cases (n = 5); patients carefully selected on the basis of their phenotype (spasticity of the lower limbs with cerebellar signs and/or cerebellar atrophy on magnetic resonance imaging/computer tomography scan and/or optic neuropathy and without other signs) (n = 24); patients with hereditary spastic paraparesis referred consecutively from attending neurologists and the national reference centre in a diagnostic setting (n = 98); and the index case of a four-generation family with autosomal dominant optic neuropathy but no spasticity linked to the SPG7 locus. We identified two SPG7 mutations in 23/134 spastic patients, 21% of the patients selected according to phenotype but only 8% of those referred directly. Our results confirm the pathogenicity of Ala510Val, which was the most frequent mutation in our series (65%) and segregated at the homozygous state with spastic paraparesis in a large family with autosomal recessive inheritance. All SPG7-positive patients tested had optic neuropathy or abnormalities revealed by optical coherence tomography, indicating that abnormalities in optical coherence tomography could be a clinical biomarker for SPG7 testing. In addition, the presence of late-onset very slowly progressive spastic gait (median age 39 years, range 18-52 years) associated with cerebellar ataxia (39%) or cerebellar atrophy (47%) constitute, with abnormal optical coherence tomography, key features pointing towards SPG7-testing. Interestingly, three relatives of patients with heterozygote SPG7 mutations had cerebellar signs and atrophy, or peripheral neuropathy, but no spasticity of the lower limbs, suggesting that SPG7 mutations at the heterozygous state might predispose to late-onset neurodegenerative disorders, mimicking autosomal dominant inheritance. Finally, a novel missense SPG7 mutation at the heterozygous state (Asp411Ala) was identified as the cause of autosomal dominant optic neuropathy in a large family, indicating that some SPG7 mutations can occasionally be dominantly inherited and be an uncommon cause of isolated optic neuropathy. Altogether, these results emphasize the clinical variability associated with SPG7 mutations, ranging from optic neuropathy to spastic paraplegia, and support the view that SPG7 screening should be carried out in both conditions.

[PMID: 23065789](#) [PubMed - indexed for MEDLINE] PMID: PMC3470714 [Available on 2013/10/1]

**22. J Perinat Med. 2013 Jan 1;41(1):107-14. doi: 10.1515/jpm-2012-0018.**

**Comparison between antenatal neurodevelopmental test and fetal Doppler in the assessment of fetal well being.**

Kurjak A, Talic A, Honemeyer U, Stanojevic M, Zalud I.

**Aims:** The primary aim of this study was to compare circulatory changes in the fetal brain under certain pathological conditions with alterations in fetal behavior. **Patients and methods:** A prospective longitudinal cohort study on fetal behavior of fetuses from singleton pregnancies between the 28th and 38th gestational week in the period from March 2009 to October 2011 was undertaken. There were 596 fetuses in the high-risk group and 273 fetuses in the low-risk group. Elevated umbilical artery Doppler pulsatility index and reduced middle cerebral artery pulsatility index obtained in the absence of fetal movements were considered abnormal. The Kurjak Antenatal Neurodevelopmental Test (KANET) was used to assess fetal behavior. **Results:** Statistically significant differences in the distribution of normal, abnormal, and borderline KANET scores between low-risk and high-risk groups were found. Furthermore, 596 fetuses from the high-risk group were subdivided into subgroups according to the risk factor. The largest proportion of abnormal KANET scores (23.9%) was in the subgroup of fetuses whose mothers had an offspring diagnosed with cerebral palsy (23.9%), followed by the proportion of borderline KANET scores in the subgroup of fetuses from febrile mothers (12.7%). Fetal behavior was significantly different between the normal group and the following subgroups of fetuses: fetal growth restriction (FGR), gestational diabetes mellitus, threatened preterm birth, antepartum hemorrhage, maternal fever, sibling with cerebral palsy, and polyhydramnios.

Conclusions: A new clinical application of the KANET test in early identification of fetuses at risk for adverse neurological outcome was demonstrated.

[PMID: 23314515](#) [PubMed - in process]

**23. J Perinatol. 2013 Jan 17. doi: 10.1038/jp.2012.167. [Epub ahead of print]**

**Neurodevelopmental outcomes of very low birth weight infants with neonatal sepsis: systematic review and meta-analysis.**

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**OBJECTIVE:** To study the impact of neonatal sepsis on the long-term neurodevelopmental outcome in very low birth weight (VLBW) infants. **STUDY DESIGN:** Systematic review and meta-analysis of observational studies comparing neurodevelopmental outcomes in VLBW infants exposed to culture-proven sepsis in the neonatal period with similar infants without sepsis. **RESULT:** Seventeen studies involving 15 331 infants were included in the meta-analysis. Sepsis in VLBW infants was associated with an increased risk of one or more long-term neurodevelopmental impairments (odds ratio (OR) 2.09; 95% confidence interval (CI) 1.65 to 2.65) including cerebral palsy (CP; OR 2.09; 95% CI 1.78 to 2.45). Heterogeneity ( $I^2=36.9\%$ ;  $P=0.06$ ) between the studies was significant and related to variations in patient characteristics, causative pathogens and follow-up methods. Sensitivity analyses based on study design, follow-up rate and year of birth were not significantly different from the overall analysis. **CONCLUSION:** The meta-analysis suggests that sepsis in VLBW infants is associated with a worse neurodevelopmental outcome including higher incidence of CP.

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[PMID: 23328927](#) [PubMed - as supplied by publisher]

**24. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;112(7 Pt 2):4-8**

**Contemporary views of the morphological basis of infant cerebral palsy [Article in Russian]**

[No authors listed]

The stages of development of infant cerebral palsy (ICP) in 300 patients during the childhood period, beginning from the neonate period, are presented in this article. Clinico-morphological analysis has been realized in 35 cases with the mortal outcome. It has been established, that combination of the following signs is the morphological basis of ICP: dysontogenetic development of the separate structures of the brain, cerebrovascular disturbances, dystrophic changes of the structural elements of the brain and in number of cases--of the focal inflammation of the brain matter, predominantly in the region of walls of the lateral ventricle of the brain. In parallel with the destructive changes in the brain the compensatory restorative processes have been also observed.

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