This free weekly bulletin lists the latest research on cerebral palsy (CP), as indexed in the NCBI, PubMed (Medline) and Entrez (GenBank) databases. These articles were identified by a search using the key term "cerebral palsy".

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Interventions

Factors Related to Physical Activity in Adults with Cerebral Palsy May Differ for Walkers and Nonwalkers.
Maltais DB, Dumas F, Boucher N, Richards CL.
From the Department of Rehabilitation, Laval University (DBM, FD, CLR) and Centre for Interdisciplinary Research in Rehabilitation and Social Integration (DBM, FD, NB, CLR), Québec City, Canada.

OBJECTIVE: To explore what factors besides walking ability, e.g., additional health problems or complications, general health, and sociodemographic status, may be related to physical activity in adults with cerebral palsy. DESIGN: We administered a questionnaire regarding sociodemographic and health-related factors of potential relevance to physical activity to 66 men (20-41 yrs) and 66 women (18-39 yrs) with various types of cerebral palsy. Data were analyzed using logistic regression. RESULTS: Use of walking as the primary means of self-transport (walking ability) was associated with a higher odds of being physically active (odds ratio = 3.75; P = 0.002). Among those who could walk, being younger and having a positive perception of health were also associated with a higher odds of being active (odds ratios of 2.6 and 3.0, respectively). This was not true among nonwalkers. For individuals who walked, inactivity was associated with an increase in the severity (during the past 3 yrs) of several additional health problems or complications. For the nonwalkers, inactivity was most clearly associated with perceived range-of-motion limitations. CONCLUSIONS: Among adults with cerebral palsy, the ability to walk, as expected, is associated with being physically active. The factors additionally related to physical activity differ between walkers and nonwalkers.

PMID: 20463567 [PubMed - as supplied by publisher]

Action planning in typically and atypically developing children (unilateral cerebral palsy).
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Radboud University Nijmegen, Donders Institute for Brain, Cognition and Behaviour, The Netherlands; Radboud University Nijmegen, Behavioural Science Institute, The Netherlands.

In the present study, we investigated the development of action planning in children with unilateral Cerebral Palsy (CP, aged 3-6 years, n=24) and an age matched control group. To investigate action planning, participants performed a sequential movement task. They had to grasp an object (a wooden play sword) and place the sword in a hole in a wooden block. Our main dependent variable was the grip type that participants used, i.e., did they adapt their initial grip choice such that they would reach a comfortable posture at the end of the action? This end-state comfort effect has been abundantly shown in research on action planning, and is taken as evidence for anticipa-
tory planning. The first aim of the study was to investigate the development of action planning in the unilateral CP group and the control group. Our hypothesis was that action planning improves with age in the control group, but not in the unilateral CP group. The results showed that planning was impaired in the unilateral CP group compared with the control group. Consistent with our hypothesis, we found an age effect in the control group, but not in the unilateral CP group. In the control group 5 and 6 years olds showed more anticipatory planning compared with the 3 and 4 years olds. The second aim of this study was to examine whether an intervention for children with unilateral CP (i.e., constrained induced movement therapy combined with bimanual training) affected action planning. The children with unilateral CP were therefore measured on the experimental task before and after an 8-week intervention period. The results showed that planning improved after the intervention. This finding suggests that action planning ability in young children with unilateral CP may be sensitive to improvement. These findings are discussed within the context of typical and atypical development of action planning and further guidelines for intervention in children with unilateral CP are given. Copyright © 2010 Elsevier Ltd. All rights reserved.

PMID: 20451346 [PubMed - as supplied by publisher]


The impact of menstruation in adolescents with disabilities related to cerebral palsy.

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Background: Information regarding menstrual difficulties for adolescents with developmental disabilities and their families is limited. Aim: To assess the impact of menstruation on adolescents with developmental disabilities and their families, and to compare this to previously reported experiences of age-matched normal girls. Methods: Families of girls aged 12-18 years with known disabilities, attending the Royal Children's Hospital, Melbourne, were recruited into a questionnaire based study evaluating issues of menstruation and associated problems, together with the consequent psychological, social and emotional impact on their families and carers. Information was sought regarding menstrual management strategies, outcome satisfaction and specific areas of family concern. Results: 103 questionnaires were completed. The average age of participating girls was 15.11 years, mean menarchal age 12.3 years. 79 girls were postmenarchal. The severity of menstrual problems was similar to a normal population. 59 (76%) were happy with the impact of menses on their social activities. More than 50% sought menstrual advice before menarche. Advice seeking strongly correlated with disability severity (p=0.01) and impact of menses on social activities (p=0.01), which in turn were highly predictive of seeking assistance (p=0.005). Carer satisfaction with current management inversely correlated with treatment seeking behaviour (p=0.034). Conclusions: Menstrual characteristics in this population are similar to those without disabilities. There is a high level of parental anxiety regarding the impact of menses, particularly when disability is severe. Medical therapies may be required but information for families is lacking. Clinicians should play a proactive and educational role with families and adolescents with disabilities.

PMID: 20457697 [PubMed - as supplied by publisher]


Quantifying the physical, social and attitudinal environment of children with cerebral palsy.

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Purpose. To develop an instrument to represent the availability of needed environmental features (EFs) in the physical, social and attitudinal environment of home, school and community for children with cerebral palsy. Method. Following a literature review and qualitative studies, the European Child Environment Questionnaire (ECEQ) was developed to capture whether EFs needed by children with cerebral palsy were available to them: 24, 24 and 12 items related to the physical, social and attitudinal environments, respectively. The ECEQ was administered to parents of 818 children with cerebral palsy aged 8-12 years, in seven European countries. A domain structure was developed using factor analysis. Results. Parents responded to 98% of items. Seven items were omitted
from statistical models as the EFs they referred to were available to most children who needed them; two items were omitted as they did not fit well into plausible domains. The final domains, based on 51 items, were: Transport, Physical - home, Physical - community, Physical - school, Social support - home, Social support - community, Attitudes - family and friends, Attitudes - teachers and therapists, Attitudes - classmates. Conclusion. ECEQ was acceptable to parents and can be used to assess both the access children with cerebral palsy have to the EFs that they need and how available individual EFs are.

PMID: 20455710 [PubMed - as supplied by publisher]


The Early Motor Repertoire of Children Born Preterm Is Associated With Intelligence at School Age.

Bruggink JL, Van Braeckel KN, Bos AF.

Division of Neonatology, Department of Pediatrics, Beatrix Children's Hospital, University Medical Center Groningen, University of Groningen, Groningen, Netherlands.

Objectives: The goal was to determine whether the quality of general movements (GMs) for preterm children had predictive value for cognitive development at school age. Methods: In this prospective cohort study, 60 preterm infants (gestational age, median: 30.0 weeks [range: 25-33 weeks]; birth weight, median: 1130 g [range: 595-1800 g]) without cerebral palsy were studied. The quality of GMs was assessed prospectively as normal or abnormal, from video recordings that were made at regular intervals until 17 weeks after term. At 7 to 11 years, intelligence was tested by using the Wechsler Intelligence Scale for Children III, Dutch version. Total IQ (TIQ), verbal IQ (VIQ), and performance IQ (PIQ) scores were calculated. Results: The median TIQ was 93 (range: 67-113), VIQ 96 (range: 68-117), and PIQ 92 (range: 65-119). Fifteen children (25%) had low TIQ scores (<85). When the quality of GMs normalized before 8 weeks after term, TIQ, VIQ, and PIQ scores were in the normal range. Consistently abnormal GMs to 8 weeks after term were associated with lower TIQ, VIQ, and PIQ scores. With correction for male gender and the educational levels of the parents, the likelihood ratio of consistently abnormal GMs for a low TIQ was 4.9 (95% confidence interval: 1.3-17.6). The model explained 22.4% of the variance. Conclusions: The quality of GMs during the early postterm period is a marker for intelligence at school age. Abnormal GMs during the early postterm period may reflect injury or developmental disruptions of brain areas involved in cognitive development.

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Using the manual ability classification system in young adults with cerebral palsy and normal intelligence.

van Meeteren J, Nieuwenhuijsen C, de Grund A, Stam HJ, Roebroeck ME.

Department of Rehabilitation Medicine, Erasmus MC, University Medical Centre, Rotterdam, The Netherlands.

Purpose. The study aimed to establish whether the manual ability classification system (MACS), a valid classification system for manual ability in children with cerebral palsy (CP), is applicable in young adults with CP and normal intelligence. Subjects. The participants (n = 83) were young adults with CP and normal intelligence and had a mean age of 19.9 years. Method. In this study, inter observer reliability of the MACS was determined. We investigated relationships between the MACS level and patient characteristics (such as the gross motor function classification system (GMFCS) level, limb distribution of the spastic paresis and educational level) and with functional activities of the upper extremity (assessed with the Melbourne assessment, the Abilhand questionnaire and the domain self-care of the functional independence measure (FIM)). Furthermore, with a linear regression analysis it was determined whether the MACS is a significant determinant of activity limitations and participation restrictions. Results. The reliability was good (intraclass correlation coefficient 0.83). The Spearman correlation coefficients with GMFCS level, limb distribution of the spastic paresis and educational level were 0.53, 0.46, and 0.26, respectively. MACS level correlated moderately with outcome measures of functional activities (correlations ranging from -0.38 to -0.55). MACS level is, in addition to the GMFCS level, an important determinant for limitations in activities and restrictions in participation. Conclusion. We conclude that the MACS is a feasible method to classify manual ability in young adults with CP and normal intelligence with a good manual ability.

Predictors of Reading Comprehension in Children with Cerebral Palsy and Typically Developing Children.

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Predictors of reading comprehension were evaluated in 41 children with cerebral palsy and 74 typically developing children between the ages of 6 and 12 years. Regression analyses were conducted to determine the relative contributions of measures of phonemic awareness, receptive vocabulary, and general reasoning to variance in reading comprehension. All three independent variables were statistically significant predictors of reading comprehension in both groups of participants. The impact of phonemic awareness on reading comprehension was moderated by age, but only in the typically developing group. Within the group with cerebral palsy, there was an indirect effect of functional expressive ability on reading comprehension, mediated by phonemic awareness. It is concluded that largely the same variables predict reading comprehension in children with cerebral palsy as in typically developing children, but that children with cerebral palsy continue to rely on phonological processing for a more protracted period of time.

PMID: 20455127 [PubMed - as supplied by publisher]


Question 1. Do Lycra garments improve function and movement in children with cerebral palsy?

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PMID: 20457706 [PubMed - in process]


Recent skin injuries in children with motor disabilities.

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OBJECTIVE: To determine the frequency of recent skin injuries in children with neuromotor disabilities and its association with disability. DESIGN: Cross-sectional study of 168 children with neuromotor disabilities aged 2-16 years. SETTING: Two outpatient child rehabilitation centres. MAIN OUTCOME MEASURES: Children were classified as unrestricted walkers, restricted walkers or wheelchair dependent. Each participant's body surface was systematically examined for recent skin injuries with the exception of the anal-genital area. RESULTS: The mean age of our sample was 7.8 (SD 3.7) years with a 3:2 male/female ratio. Overall, 64% had cerebral palsy, 17% a neuromuscular disease and 19% other motor disabilities. Participants had on average 5.3 (SD 4.5) recent skin injuries (max 19), of which 2.5 were bruises (SD 3.3, max 16), 2.4 were abrasions, scratches or cuts (SD 3.0, max 16) and 0.4 were pressure lesions (SD 0.8, max 4). There was a significant decrease in the frequency of recent skin injuries and of bruises with increasing severity of motor disability. Most of this variation was accounted for by injuries to the lower limbs. There were no significant effects of gender, learning disabilities or other comorbidities. CONCLUSIONS: Children with neuromotor disabilities present a progressive reduction in the number of skin injuries with decreasing mobility. Therefore, recent skin injuries in this population which are unusual by their number, appearance or distribution, should raise at least the same level of suspicion for physical abuse as in children without disabilities.

Significant sleep dysregulation in a toddler with developmental delay.

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CASE: Derrick's parents made an appointment with a new pediatrician for a second opinion about disordered sleep. Now 22-months old, he was evaluated at 18 months of age for developmental delay when he was found to have “a regulatory disorder associated with delays in language and motor development, hypotonia and significant sleep problems.” The parents are now most concerned about his sleeping pattern. Prolonged sleep onset and frequent night waking occur each night since 6-months of age. These problems are more severe in the past few months when he awakes screaming and cannot be settled. The awakening episodes occur 2 to 4 times each night when "he screams and thrashes his body for up to an hour." Daytime tantrums increased. After the parents read a book about sleep in young children, they provided a calm atmosphere at bedtime including a dark room and singing a quiet lullaby. When these changes did not alter sleep, they purchased a vibrating mattress which was also unsuccessful. Derrick was born full term after an uncomplicated prenatal and perinatal course. He sat at 10 months, crawled at 12 months, and walked at 18 months. He currently drinks from a sippy cup and he can use a utensil to eat. He has few words saying only "no" and "mama" in the past month. Imitation of some words occurred recently. He has responded to simple directions in the past 2 months. Derrick passed the newborn audiology screen. He does not have difficulty swallowing and he does not drool. He plays with many different toys and he plays in parallel with his older brother who also experienced delays in motor and language development. His brother is now doing very well in school. There is no family history of cognitive delay, seizure disorder, cerebral palsy, early developmental delay (other than the brother) or a significant sleep problem. Physical examination: head circumference, length and weight (75th percentile). He had mild generalized hypotonia, mild weakness, 2+ symmetrical deep tendon reflexes, and absence of ankle clonus. His gait was slightly wide based, steady, and without a limp. Neither ataxia nor drooling was observed. He was easily engaged in play with the examiner without evidence of irritability. The remainder of the examination was normal.


Efficacy and safety of NT 201 for upper limb spasticity of various etiologies - a randomized parallel-group study.

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Objective - To assess efficacy and safety of two dilutions of botulinum neurotoxin type A NT 201 (Xeomin((R))) in patients with upper limb spasticity of diverse etiology. Methods - Changes in functional disability and muscle tone from baseline to week 4 after NT 201 treatment. Results - One hundred ninety-two patients with stroke, brain injury, multiple sclerosis, or cerebral palsy were randomized to either 50 or 20 U/ml NT 201 dilutions. The maximum total NT 201 dose was 495 units. Four weeks post-injection, a >/= 1-point reduction was observed on the Disability Assessment Scale in 57.1%, and on the Ashworth scale in >/= 62.2% of patients. The 20 U/ml NT 201 dilution was non-inferior to the 50 U/ml NT 201 dilution. Global improvement was rated high by patients (80.2%) and investigators (89.0%). Conclusions - NT 201 improved functional disability and muscle tone and was well tolerated in patients with upper limb spasticity of diverse etiology in both dilutions.

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PMID: 20453583 [PubMed - in process]

PMID: 20456248 [PubMed - as supplied by publisher]

Developmental and degenerative features in a complicated spastic paraplegia.


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OBJECTIVE: We sought to explore the genetic and molecular causes of Troyer syndrome, one of several complicated hereditary spastic paraplegias (HSPs). Troyer syndrome had been thought to be restricted to the Amish; however, we identified 2 Omani families with HSP, short stature, dysarthria and developmental delay-core features of Troyer syndrome-and a novel mutation in the SPG20 gene, which is also mutated in the Amish. In addition, we analyzed SPG20 expression throughout development to infer how disruption of this gene might generate the constellation of developmental and degenerative Troyer syndrome phenotypes. METHODS: Clinical characterization of 2 non-Amish families with Troyer syndrome was followed by linkage and sequencing analysis. Quantitative polymerase chain reaction and in situ hybridization analysis of SPG20 expression were carried out in embryonic and adult human and mouse tissue. RESULTS: Two Omani families carrying a novel SPG20 mutation displayed clinical features remarkably similar to the Amish patients with Troyer syndrome. SPG20 mRNA is expressed broadly but at low relative levels in the adult brain; however, it is robustly and specifically expressed in the limbs, face, and brain during early morphogenesis. INTERPRETATION: Null mutations in SPG20 cause Troyer syndrome, a specific clinical entity with developmental and degenerative features. Maximal expression of SPG20 in the limb buds and forebrain during embryogenesis may explain the developmental origin of the skeletal and cognitive defects observed in this disorder.

PMID: 20437587 [PubMed - indexed for MEDLINE]


Physical fitness, everyday physical activity, and fatigue in ambulatory adults with bilateral spastic cerebral palsy.


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This study assessed physical fitness and its relationships with everyday physical activity (PA) and fatigue in cerebral palsy (CP). Participants were 42 adults with ambulatory bilateral spastic CP (mean age 36.4 +/- 5.8 years; 69% males; 81% with good gross motor functioning). Progressive maximal aerobic cycle tests determined VO(2peak) (L/min). Objective levels of everyday PA were measured with accelerometry and self-reported levels of everyday PA with the Physical Activity Scale for Individuals with Physical Disabilities. Fatigue was assessed with the Fatigue Severity Scale. The average aerobic capacity of adults with CP was 77% of Dutch reference values. Participants were physically active during 124 min/day (85% of Dutch reference values), and half experienced fatigue. In women, lower physical fitness was related to lower self-reported levels of PA (R(p)=0.61, P=0.03), and in men to higher levels of fatigue (R(p)=-0.37, P=0.05). Other relationships were not significant. Results suggest that ambulatory adults with CP have low levels of physical fitness, are less physically active than able-bodied age mates and often experience fatigue. We found little evidence for relationships between the level of physical fitness and everyday PA or fatigue.

PMID: 20459469 [PubMed - as supplied by publisher]

Effect of penetrative needling of otopoints combined with body acupuncture on limb myodynamia and neurofunction in patients with acute cerebral infarction [Article in Chinese]

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OBJECTIVE: To compare the therapeutic effects of different acupuncture methods in improving the myodynamia and neurofunction of patients with acute cerebral infarction (ACI). METHODS: A total of 90 ACI patients were randomized into ear-acupuncture, scalp-acupuncture and body-acupuncture groups, with 30 cases in each. For patients of ear-acupuncture group, the main otopoints used for penetrative needling were Zhen(MA-AT)-Nie(MA-AT)-E(MA-AT) on the affected side in combination with Jian(MA-SF 4)-Suogu(MA-SF 5) and Zhou(MA-SF 3)-Wan(MA-SF 2)-Zhi(MA-SF 1) for upper-limb paralysis, and with Tun (MA-AH 5)-Zuogushenjing(MA-AH 6), Kuan(MA-AH 4)-Xi(MA-AH 3), and Xi(MA-AH 3)-Huai(MA-AH 2)-Zhi(MA-AH 1) for lower-limb paralysis, and body acupoints as Ji-anyu(LI 15), Hegu(LI 4), Huantiao(GB 30), Taixi(KI 3), etc. For patients of scalp-acupuncture group, scalp-points used were Dingnie Qianxieixian(MS 6) and Dingnie Houxieixian(MS 7) on the healthy side, and combined with body acupoints (the same as those mentioned above). For patients of body-acupuncture group, only body acupoints were used. The treatment was given once daily for 14 days. RESULTS: Comparison among 3 groups showed that the increased myodynamia of both upper and lower limbs in ear-acupuncture and scalp-acupuncture groups was significantly superior to that of body-acupuncture group (P < 0.01). The neurofunctional deficit scores of ear-acupuncture and scalp-acupuncture groups were significantly lower than that of body-acupuncture group (P < 0.01) after the treatment. In comparison with pre-acupuncture, the neurofunctional deficit scores of the 3 groups lessened considerably after the treatment (P < 0.01). Of the 30 cases in each of earacupuncture, scalp-acupuncture and body-acupuncture groups, 0, 1 and 0 were cured basically, 21, 18 and 4 experienced marked improvement, 9, 11 and 20 were improved, 0, 0 and 6 failed, respectively. The therapeutic effects of ear-acupuncture and scalp-acupuncture were obviously superior to that of simple body acupuncture. CONCLUSION: Acupuncture can effectively improve ACI patients' clinical symptoms and the therapeutic effect of ear-acupuncture and scalp-acupuncture was superior to that of simple body acupuncture.

PMID: 20458909 [PubMed - in process]


Clinical profile and treatment of infantile spasms using vigabatrin and ACTH--a developing country perspective.

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BACKGROUND: Infantile spasms represent a serious epileptic syndrome that occurs in the early infantile age. ACTH and Vigabatrin are actively investigated drugs in its treatment. This study describes the comparison of their efficacy in a large series of patients with infantile spasms from Pakistan. METHODS: All patients with infantile spasms who presented to Aga Khan University Hospital, Karachi, Pakistan from January, 2006 to April, 2008 were included in this study. Inclusion criteria were clinical symptoms of infantile spasms, hypsarrythmia or modified hypsarrythmia on electroencephalography, at least six months of follow-up period and receipt of any of the two drugs mentioned above. The type of drug distribution was random according to the availability, cost and ease of administration. RESULTS: Fifty six cases fulfilled the inclusion criteria. 62.5% were males. Mean age at onset of seizures was 5 +/- 1.4 months. Fifty two (92.8%) patients demonstrated hypsarrythmia on electroencephalography. 64.3% cases were identified as symptomatic while 19.6% were cryptogenic and 16.1% were idiopathic. Eighteen patients received ACTH while 38 patients received Vigabatrin as first line therapy. Initial response to first line therapy was similar (50% for ACTH and 55.3% for Vigabatrin). Overall, the symptomatic and idiopathic groups responded better to Vigabatrin. The relapse rate was higher for ACTH as compared to Vigabatrin (55.5% vs. 33.3%) when considering the first line therapy. Four patients evolved to Lennox-Gastaut variant; all of these patients had initially received
Vigabatrin and then ACTH. CONCLUSION: Vigabatrin and ACTH showed no significant difference in the initial treatment of infantile spasms. However, patients receiving ACTH were 1.2 times more likely to relapse as compared to the patients receiving Vigabatrin when considering monotherapy. We suggest that Vigabatrin should be the initial drug of choice in patients presenting with infantile spasms. However, larger studies from developing countries are required to validate the therapeutic trends observed in this study.

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PURPOSE: This study explores post-operative family situation, rehabilitation and interdisciplinary cooperation for ambulant children with cerebral palsy (CP), after multilevel surgery. METHODS: Eight ambulant spastic children with varied severity of CP and their parents were included. Qualitative, semi-structured interviews were carried out separately with children and parents. RESULTS: Children experiencing a low degree of post-operative pain were satisfied due to improved strength and ambulation, leading to increased social participation. A few experienced severe pain and modest physical improvement. Most families experienced a lack of information and communication between rehabilitation levels. Rehabilitation was considered strenuous because of complex and intense training programmes. Schools were mostly responsive to children's extra post-operative needs, but some examples of serious neglect and bullying occurred. CONCLUSION: Results imply the need for systematic securing of interdisciplinary knowledge transfer regionally and locally by the university hospital, aiming at empowering families and health professionals involved in this complex rehabilitation.

PMID: 20450468 [PubMed - in process]

Epidemiology / Aetiology / Diagnosis & Early Treatment

Please note: This is not yet a comprehensive outline of cerebral palsy prevention literature. It is expected that more research will be included when the search terms are expanded to include key terms other than "cerebral palsy". It is a work-in-progress and it will be expanded in coming months.


Strength of association between umbilical cord pH and perinatal and long term outcomes: systematic review and meta-analysis.

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Comment in:

BMJ. 2010;340:c1720.

OBJECTIVE: To evaluate the association between umbilical cord pH at birth and long term outcomes. DESIGN: Systematic review and meta-analysis. DATA SOURCES: Medline (1966-August 2008), Embase (1980-August 2008), the Cochrane Library (2008 issue 8), and Medion, without language restrictions; reference lists of selected
articles; and contact with authors. STUDY SELECTION: Studies in which cord pH at birth was compared with any neonatal or long term outcome. Cohort and case-control designs were included. RESULTS: 51 articles totalling 481,753 infants met the selection criteria. Studies varied in design, quality, outcome definition, and results. Meta-analysis carried out within predefined groups showed that low arterial cord pH was significantly associated with neonatal mortality (odds ratio 16.9, 95% confidence interval 9.7 to 29.5, I(2)=0%), hypoxic ischaemic encephalopathy (13.8, 6.6 to 28.9, I(2)=0%), intraventricular haemorrhage or periventricular leucomalacia (2.9, 2.1 to 4.1, I(2)=0%), and cerebral palsy (2.3, 1.3 to 4.2, I(2)=0%). CONCLUSIONS: Low arterial cord pH showed strong, consistent, and temporal associations with clinically important neonatal outcomes that are biologically plausible. These data can be used to inform clinical management and justify the use of arterial cord pH as an important outcome measure alongside neonatal morbidity and mortality in obstetric trials.

PMID: 20466789 [PubMed - in process]


Dominant mutations in the cation channel gene transient receptor potential vanilloid 4 cause an unusual spectrum of neuropathies.


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Hereditary neuropathies form a heterogeneous group of disorders for which over 40 causal genes have been identified to date. Recently, dominant mutations in the transient receptor potential vanilloid 4 gene were found to be associated with three distinct neuromuscular phenotypes: hereditary motor and sensory neuropathy 2C, scapuloperoneal spinal muscular atrophy and congenital distal spinal muscular atrophy. Transient receptor potential vanilloid 4 encodes a cation channel previously implicated in several types of dominantly inherited bone dysplasia syndromes. We performed DNA sequencing of the coding regions of transient receptor potential vanilloid 4 in a cohort of 145 patients with various types of hereditary neuropathy and identified five different heterozygous missense mutations in eight unrelated families. One mutation arose de novo in an isolated patient, and the remainder segregated in families. Two of the mutations were recurrent in unrelated families. Four mutations in transient receptor potential vanilloid 4 targeted conserved arginine residues in the ankyrin repeat domain, which is believed to be important in protein-protein interactions. Striking phenotypic variability between and within families was observed. The majority of patients displayed a predominantly, or pure, motor neuropathy with axonal characteristics observed on electrophysiological testing. The age of onset varied widely, ranging from congenital to late adulthood onset. Various combinations of additional features were present in most patients including vocal fold paralysis, scapular weakness, contractures and hearing loss. We identified six asymptomatic mutation carriers, indicating reduced penetrance of the transient receptor potential vanilloid 4 defects. This finding is relatively unusual in the context of hereditary neuropathies and has important implications for diagnostic testing and genetic counselling.

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Impact of chorioamnionitis and preeclampsia on neurodevelopmental outcome in preterm infants below 32 weeks gestational age.

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Aim: Intrauterine conditions may interfere with fetal brain development. We compared the neurodevelopmental outcome between infants <32 weeks gestational age after maternal preeclampsia or chorioamnionitis and controls. Methods: Case-control study on infants with maternal preeclampsia, chorioamnionitis and controls (each n = 33) matched for gestational age. Neurodevelopment at two years was assessed with the Bayley Scales of Infant Development II. Results: Ninety-nine infants were included with a median gestational age of 29 weeks (range 25-32). Me-
dian mental developmental index (MDI) was 96 in the control, 90 in the chorioamnionitis and 86 in the preeclampsia group. Preeclampsia infants had a lower MDI compared with the control group (univariate p = 0.021, multivariate p = 0.183) and with the chorioamnionitis group (univariate p = 0.242; multivariate p = 0.027). Median psychomotor index was 80.5 in the control, 80 in the preeclampsia and 85 in the chorioamnionitis group, and was not different between these three groups (p > 0.05). Chorioamnionitis or preeclampsia exposure was not associated with major neurodevelopmental impairments (cerebral palsy, MDI<70, PDI<70).

Conclusion: The results of this preliminary study suggest that preeclampsia and chorioamnionitis play a relatively minor role among risk factors for adverse neurodevelopment outcome. Postnatal factors such as ventilation and bronchopulmonary dysplasia may have a greater impact on neurodevelopmental outcome.

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Cerebral palsy in children born after in vitro fertilization. Is the risk decreasing?

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BACKGROUND: Infants born after in vitro fertilization (IVF) differ from spontaneously conceived infants in a number of aspects which could increase the risk for future cerebral palsy (CP), e.g., multiple births, preterm births, neonatal complications. AIMS: To follow up children conceived by IVF with respect to risk for CP. METHODS: Infants born after IVF were identified from all IVF clinics in Sweden 1982-2007. Perinatal characteristics were obtained by linkage with the Medical Birth Register. The presence of CP in children born after IVF and in other children was identified from the Patient Register which contains diagnoses given at hospitalizations or specialist outpatient clinics. The risk for CP after IVF was studied after adjustment for year of birth, maternal age, parity, and smoking, all factors which co-vary both with IVF and with CP. Stratification was made for singletons and multiple births and for various neonatal outcomes. RESULTS: The adjusted odds ratio for CP after IVF was 1.81 (95% confidence interval, 95% CI 1.52-2.13), lower and not statistically significant when singletons or when unlike-sexed twins were analyzed. Stratification for various neonatal characteristics also reduced odds ratios to non-significant levels. For the last few years of the study (2004-2007) when the twinning rate after IVF was <10%, the odds ratio for CP was 0.97 (95% CI 0.57-1.66). CONCLUSIONS: The moderately increased risk for CP was most likely a consequence of an increased risk of neonatal morbidity, notably associated with multiple births. Copyright © 2010 European Paediatric Neurology Society. Published by Elsevier Ltd. All rights reserved.

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Dexamethasone Treatment after the First Week of Life for Bronchopulmonary Dysplasia in Preterm Infants: A Systematic Review.

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Background: Dexamethasone has powerful anti-inflammatory effects and has been used to treat established bronchopulmonary dysplasia (BPD), but it is uncertain whether the benefits outweigh the risks of treatment. Objectives: To determine the effect of late (>7 days) postnatal dexamethasone treatment compared with control (placebo or nothing) to prevent or treat BPD in the preterm infant. Methods: Randomised controlled trials (RCTs) of late postnatal dexamethasone therapy to treat or prevent BPD were sought using methods of the Cochrane Collaboration. Data regarding clinical outcomes including mortality, BPD, death or BPD, complications during the primary hospitalisation, and long-term outcome were ed and analysed using RevMan 5. Results: 19 RCTs enrolling 1,345 participants were eligible for this review. Late dexamethasone treatment reduced neonatal mortality, but not later mortality. Benefits of late dexamethasone included reductions in failure to extubate, BPD and the combined outcome of death or BPD. There were clear short-term complications, including hyperglycaemia and hypertension, but not in-
testinal perforation. Trends of an increase in cerebral palsy or abnormal neurological examination were partly offset by a trend in the opposite direction in death before late follow-up. Conclusions: The benefits of late dexamethasone may not outweigh actual or potential adverse effects. Given the evidence of both benefits and harms of treatment, and the limitations of the evidence at present, it appears prudent to reserve the use of late dexamethasone to infants who cannot be weaned from mechanical ventilation, and to minimise the dose and duration of any course of treatment. Copyright © 2010 S. Karger AG, Basel.

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Congenital cytomegalovirus infection; the impact of cerebral cortical malformations.

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Aim: Cytomegalovirus has been suggested to have a teratogenous influence during the migration of neural cells from the ventricular zones to the cortex during the gestational period. The aim of this study was to investigate the prevalence of congenital cytomegalovirus infections in a cohort of children with neurological disability and cerebral cortical malformations recognised by neuroimaging. Methods: Twenty-six children with neurological disability and cerebral cortical malformations were investigated retrospectively for congenital cytomegalovirus infection by analysing the dried blood spot samples for cytomegalovirus deoxynucleic acid using qualitative polymerase chain reaction. Results: CMV DNA in the dried blood spot samples was found in four out of 26 children. Two of these four had severe disabilities with mental retardation, autism, spastic cerebral palsy, epilepsy and deafness. A third child had epilepsy and unilateral cerebral palsy, while the fourth had a mild motor coordination dysfunction and hearing deficit. Conclusion: In our study the number of congenital cytomegalovirus infections in children with cerebral cortical malformations was higher (4/26) than expected with reference to the birth prevalence (0.2-0.5%) of congenital cytomegalovirus infection in Sweden. We thus conclude that congenital cytomegalovirus infection, should be considered in children with cortical malformations of unknown origin.

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