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Interventions and Management

1. Scand J Occup Ther. 2014 Mar 28. [Epub ahead of print]

Hand function in the play behavior of children with cerebral palsy.

Pfeifer LI1, Santos TR, Silva DB, Panúncio Pinto MP, Caldas CA, Santos JL.

Objective: The aim of this study was to evaluate the influence of hand function on the occupational performance of playing for children with cerebral palsy. **Methods.** Twenty children with cerebral palsy took part in the study, of both genders, aged 3-10 years, with motor abilities ranging from I to V and manual abilities from II to IV. Data were collected using the Assessment of Ludic Behaviour before and after botulinum toxin was applied in the upper limbs to reduce spasticity. **Results:** Significant differences were found between the scores before and after the application of botulinum toxin in relation to the total Assessment of Ludic Behaviour score ($p < 0.001$), as well as in basic ludic interest ($p = 0.003$), basic ludic ability ($p < 0.001$), ludic attitude ($p = 0.008$), and communication of needs and feelings ($p = 0.025$), except for general level of interest ($p = 0.957$). **Conclusions:** The reduction of spasticity permits better handling of the play materials, which promotes the children's involvement in play situations.

[PMID: 24678715](https://pubmed.ncbi.nlm.nih.gov/24678715/) [PubMed - as supplied by publisher]

2. J Hand Surg Eur Vol. 2014 Apr 1. [Epub ahead of print]

The role of arthrodesis of the wrist in spastic disorders.

Neuhaus V1, Kadzielski JJ, Mudgal CS.

We investigated the functional and radiographic outcome of wrist arthrodesis in 11 adults with spastic wrist deformities, carried out by one surgeon between 2003 and 2012. The underlying cause of spasticity was a cerebrovascular insult in five, traumatic brain injury in four, and cerebral palsy in two patients. A dorsal plate and local bone graft was used in all patients. The mean radiographic flexion deformity significantly improved from 67° pre-operatively to 4° of dorsal angulation post-operatively. Thumb-in-palm deformity was more pronounced in three patients after the operation. The functional House score improved in all patients an average of two levels (range 1-3). Level of evidence: IV (Case series).

[PMID: 24692187](https://pubmed.ncbi.nlm.nih.gov/24692187/) [PubMed - as supplied by publisher]

3. Aust Occup Ther J. 2014 Apr;61(2):126-7. doi: 10.1111/1440-1630.12109.

No differences were observed between six months of context- versus child-focussed intervention for young children with cerebral palsy on self-care, mobility, range-of-motion or participation.

Wallen M1, Majnemer A.

[PMID: 24689923](#) [PubMed - in process]

4. Res Dev Disabil. 2014 Mar 25. pii: S0891-4222(14)00079-1. doi: 10.1016/j.ridd.2014.02.011. [Epub ahead of print]

Gait characteristics in children and adolescents with cerebral palsy assessed with a trunk-worn accelerometer.

Saether R1, Helbostad JL2, Adde L3, Brændvik S4, Lydersen S5, Vik T6.

This study aimed to investigate gait characteristics reflecting balance and progression in children and adolescents with cerebral palsy (CP) compared with typically developing (TD) children. Gait characteristics variables representing aspects of balance were trunk acceleration, interstride regularity and asymmetry of accelerations while gait characteristics representing progression were gait speed, cadence, step time and step length. Children in the age range 5-18 years (mean age 11.1 years) with spastic CP (n=41) and a gross motor function corresponding to GMFCS I-III and children with TD (n=29) were included. The children walked back and forth along a 5m pathway with a tri-axial accelerometer worn on the lower back to allow assessment of their gait characteristics. Data were recorded along the anteroposterior (AP), mediolateral (ML), and vertical (V) axes. To assess the magnitude of potential differences in gait characteristics, standard deviation scores were calculated, using TD children as reference. Gait parameters related to balance, such as AP, ML, and V accelerations, were higher in the children with CP (z-scores between 0.4 and 0.7) and increased with increasing GMFCS levels. The differences in accelerations in the AP and V directions increased between children with CP and TD children with increasing speed. Also asymmetry in trunk accelerations differed significantly between the two groups in all three directions (z-scores between 0.8 and 1.8 higher in the CP group), while interstride regularity differed only slightly between children with CP and TD children, and only in the AP direction. Gait characteristics also differed between children with the spastic subtypes unilateral and bilateral CP, for accelerations and asymmetry in the AP and ML directions. Our results showed significant differences in gait characteristics between children with CP and TD children. The differences may be more related to balance than progression, and these problems seem to rise with increasing gross motor impairment and speed.

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[PMID: 24679701](#) [PubMed - as supplied by publisher]

5. J Head Trauma Rehabil. 2014 Apr 1. [Epub ahead of print]

Classification of Gait Disorders Following Traumatic Brain Injury.

Williams G1, Lai D, Schache A, Morris ME.

OBJECTIVE: To determine the extent to which gait disorders associated with traumatic brain injury (TBI) are able to be classified into clinically relevant and distinct subgroups. **DESIGN:** Cross-sectional cohort study comprising people with TBI receiving physiotherapy for mobility limitations. **PARTICIPANTS:** One hundred two people with TBI. **OUTCOME MEASURES:** The taxonomic framework for gait disorders following TBI was devised on the basis of a framework previously developed for people with cerebral palsy. Participants with TBI who were receiving therapy for mobility problems were assessed using 3-dimensional gait analysis. Pelvis and bilateral lower limb kinematic data were recorded using a VICON motion analysis system while each participant walked at a self-selected speed. Five trials of data were collected for each participant. Multiclass support vector machine models were developed to systematically and automatically ascertain the clinical classification. **RESULTS:** The statistical features derived from the major joint angles from unaffected limbs contributed to the best classification accuracy of 82.35% (84 of the 102 subjects). Features from the affected limb resulted in a classification accuracy of 76.47% (78 of 102 subjects).

CONCLUSIONS: Despite considerable variability in gait disorders following TBI, we were able to generate a clinical classification system on the basis of 6 distinct subgroups of gait deviations. Statistical features related to the motion of the pelvis, hip, knee, and ankle on the less affected leg were able to accurately classify 82% of people with TBI-related gait disorders using a multiclass support vector machine framework.

[PMID: 24695264](#) [PubMed - as supplied by publisher]

6. Orthop Traumatol Surg Res. 2014 Mar 27. pii: S1877-0568(14)00040-1. doi: 10.1016/j.otsr.2014.01.004. [Epub ahead of print]

Percutaneous pelvic osteotomy in non-ambulatory cerebral palsy patients.

Canavese F1, De Coulon G2.

The aim of this study was to describe the surgical technique of and indications for percutaneous pelvic osteotomy in patients with severe cerebral palsy. Forty non-ambulatory children and adolescents (47 hips) were consecutively treated with percutaneous pelvic osteotomy. The mean preoperative Reimers' migration percentage improved from 66.2% to 4.9% at the final follow-up. The mean preoperative acetabular angle (AA) improved from 32.4° to 13.2° at last follow-up. Percutaneous pelvic osteotomy is a less invasive surgical approach and appears to be a valid option with similar outcomes to standard techniques. This method results in less muscle stripping and blood loss and a shorter operating time.

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7. Phys Ther. 2014 Apr 3. [Epub ahead of print]

Reduced Moderate-to-Vigorous Physical Activity and Increased Sedentary Behavior Is Associated With Elevated Blood Pressure Values in Children With Cerebral Palsy.

Ryan JM1, Hensey O, McLoughlin B, Lyons A, Gormley J.

Background: Children with cerebral palsy (CP) participate in reduced levels of physical activity and spend increased time in sedentary behavior. The effect of this on their cardiometabolic health has not been investigated. **Objectives:** The purpose of this study was to investigate the prevalence of overweight/obesity and elevated blood pressure (BP) among a cohort of ambulatory children with CP. Secondly, the aim was to investigate the association between physical activity, sedentary behavior, overweight/obesity and BP in children with CP. **Study design:** This was a cross-sectional study of 90 ambulatory children, age 6 to 17 years, with CP. **Methods:** Body mass index (BMI), waist circumference, waist-height ratio and BP were measured on one occasion. Habitual physical activity was measured by accelerometry over 7 days. **Results:** The prevalence of overweight/obesity in the cohort was 18.9%. Twenty-two percent of children had BP values within the hypertensive or pre-hypertensive range. Systolic BP was positively associated with waist circumference ($\beta = 0.324$, $p < 0.05$) and BMI ($\beta = 0.249$, $p < 0.05$). Elevated BP values were associated with reduced time in moderate-to-vigorous activity, vigorous activity and total activity, and increased time in sedentary behavior. The strongest association was observed between elevated BP and vigorous activity alone (odds ratio, 0.61; 95% confidence interval, 0.37-0.99; $p < 0.05$). **Limitations:** A convenience sample was recruited for this study and it is possible that this resulted in selection bias. **Conclusions:** Despite the relatively low prevalence of overweight/obesity, a relatively high proportion of children with CP had elevated BP values. Reducing sedentary behavior and increasing habitual physical activity, particularly vigorous activity, should be a primary aim of rehabilitation in order to reduce cardiometabolic disease risk in this population.

[PMID: 24700137](#) [PubMed - as supplied by publisher]

8. *Res Dev Disabil.* 2014 Mar 29;35(6):1301-1309. doi: 10.1016/j.ridd.2014.03.028. [Epub ahead of print]

Systematic review of the relationship between habitual physical activity and motor capacity in children with cerebral palsy.

Keawutan P1, Bell K2, Davies PS3, Boyd RN4.

Habitual physical activity (HPA) has many benefits for general health. Motor capacity in children with cerebral palsy (CP) can impact on their HPA. This study aimed to systematically review the available literature on the relationship between HPA and motor capacity in children with CP aged 3-12 years for all gross motor functional abilities (GMFCS I-V) compared to typically developing children. Five electronic databases (Pubmed, Cochrane, Embase, Cinahl and Web of Science from 1989 to November, 2013) were searched using keywords "children with cerebral palsy", "physical activity", "motor capacity" and "motor function" including their synonyms and MeSH terms. Studies were included if they (i) were conducted in children with CP aged between 3 and 12 years, (ii) assessed HPA or time spent sedentary, (iii) assessed motor capacity in order to evaluate the relationship between HPA and motor capacity. All articles retrieved were reviewed by two independent reviewers and discussed until they reached consensus. Study quality of reporting was evaluated using the Strengthening the Reporting of Observational Studies in Epidemiology (STROBE) criteria. Search results identified 864 articles but after review of the title and abstract only 21 articles warranted closer consideration. Ten articles met the strict inclusion criteria as nine articles did not assess HPA and two were conference abstracts. Study quality assessment (STROBE) found nine articles were good quality ($\geq 60\%$) and one was poor quality (55.9%). Participants were mean age 8.4 (SD=2.1) years (range 2-17 years) and included children at all GMFCS levels (3 studies), while seven studies only recruited GMFCS level I-III. HPA measurements were either subjective (Activity Scale for Kids, Dutch Questionnaire of Participation in physical activity and assessment of participation in physical education at school and regular physical activity in leisure time) or objective (StepWatch® and ActiGraph®7164). Nine studies found that motor capacity was directly associated with HPA, HPA in children with CP with high functional level (GMFCS I) was higher than those with lower functional levels (GMFCS III-V); while one study reported no relationship between HPA and GMFCS level (HPA was measured by questionnaire, a potential limitation). Further studies are required to further elucidate HPA levels (active, sedentary behavior) according to objective motor capacity measures, age and gender to inform healthy lifestyle behavior (active/sedentary) in children with CP.

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9. *PLoS One.* 2014 Mar 28;9(3):e93318. doi: 10.1371/journal.pone.0093318. eCollection 2014.

Virtual reality therapy for adults post-stroke: a systematic review and meta-analysis exploring virtual environments and commercial games in therapy.

Lohse KR1, Hilderman CG2, Cheung KL2, Tatla S3, Van der Loos HF4.

BACKGROUND: The objective of this analysis was to systematically review the evidence for virtual reality (VR) therapy in an adult post-stroke population in both custom built virtual environments (VE) and commercially available gaming systems (CG). **METHODS:** MEDLINE, CINAHL, EMBASE, ERIC, PSYCInfo, DARE, PEDro, Cochrane Central Register of Controlled Trials, and Cochrane Database of Systematic Reviews were systematically searched from the earliest available date until April 4, 2013. Controlled trials that compared VR to conventional therapy were included. Population criteria included adults (>18) post-stroke, excluding children, cerebral palsy, and other neurological disorders. Included studies were reported in English. Quality of studies was assessed with the Physiotherapy Evidence Database Scale (PEDro). **RESULTS:** Twenty-six studies met the inclusion criteria. For body function outcomes, there was a significant benefit of VR therapy compared to conventional therapy controls, $G = 0.48$, 95% CI=[0.27, 0.70], and no significant difference between VE and CG interventions ($P=0.38$). For activity outcomes, there was a significant benefit of VR therapy, $G=0.58$, 95% CI=[0.32, 0.85], and no significant difference between VE and CG interventions ($P=0.66$). For participation outcomes, the overall effect size was $G=0.56$, 95% CI=[0.02, 1.10]. All participation outcomes came from VE studies. **DISCUSSION:** VR rehabilitation moderately improves outcomes compared to conventional therapy in adults post-stroke. Current CG interventions have been too few and too small to assess potential benefits of CG. Future research in this area should aim to clearly define conventional therapy, report on participation measures, consider motivational components of therapy, and investigate commercially available systems in larger RCTs.

TRIAL REGISTRATION: Prospero CRD42013004338.

[PMID: 24681826](#) [PubMed - in process] PMID: PMC3969329 Free PMC Article

10. Indian J Pediatr. 2014 Apr 3. [Epub ahead of print]

Indian Children with Developmental Disabilities: Early versus Late Referral for Intervention.

Juneja M1, Jain R, Chakrabarty B, Mishra D, Saboo P.

OBJECTIVE: To study the age at referral, of children with neurodevelopmental disabilities to Child Development and Early Intervention Clinic and compare the neuromorbidity and socio-economic profile of the early and late presenters. **METHODS:** This retrospective observational study was conducted at Child Development and Early Intervention Clinic (CDEIC) located in Northern India. Case records of children enrolled at CDEIC in last 5 y; with neurodevelopmental disabilities namely Mental Retardation/Global Developmental Delay, Cerebral Palsy, hearing and vision impairment were separated and studied. **RESULTS:** Two thousand and twenty cases were included in this study. 62.8 % presented before 3 y of age (early presenters) and 37.1 % presented at 3 y or more (late presenters). There was no difference in the overall rates and severity of mental retardation in early and late presenters. The proportion of children with quadriparetic cerebral palsy, hearing impairment, vision impairment and multiple disabilities was significantly more in early presenters. The early presenters had better parental education status, less number of siblings, better immunization status and more were delivered at a hospital and residing in urban areas. **CONCLUSIONS:** Large numbers of children with neurodevelopmental disabilities are referred late for intervention services, leading to loss of opportunity for early intervention. Children with purely mental disability are the ones, most likely to be referred late. Socio-economic differences are significantly contributing to these delayed referrals.

[PMID: 24691576](#) [PubMed - as supplied by publisher]

11. Nutr Hosp. 2014 Apr 1;29(n04):838-843.

Intensive nutritional support improves the nutritional status and body composition in severely malnourished children with cerebral palsy.

García-Contreras AA1, Vásquez-Garibay EM2, Romero-Velarde E3, Ibarra-Gutiérrez AI4, Troyo-Sanromán R5, Sandoval-Montes IE5.

Objective: To demonstrate that a nutritional support intervention, via naso-enteral tube-feeding or gastrostomy, has a significant impact on the nutritional status and body composition in severely malnourished children with cerebral palsy spastic quadriplegia **Methods:** Thirteen patients with moderate/severe malnutrition and cerebral palsy spastic quadriplegia who were fed via naso-enteral tube-feeding or gastrostomy were included in a cohort study. Anthropometric measurements and estimated body composition by bioelectric impedance analysis were obtained. ANOVA and Wilcoxon tests were used. **Results:** During the four weeks of nutritional recovery, an average weight increase of 2700 g was achieved. There were significant increases in anthropometric indicators, including BMI and weight/length ($p < 0.01$). The increase in arm fat area was significantly higher than the increase in arm muscle area (104.5 vs 17.5%). **Conclusion:** Intensive nutritional support for four weeks had a significant effect on the nutritional status and body composition of severe and moderately malnourished children with cerebral palsy spastic quadriplegia.

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12. Indian J Anaesth. 2014 Jan;58(1):89-90. doi: 10.4103/0019-5049.126841.

Dexmedetomidine to wean patient of severe kyphoscoliosis with cerebral palsy in intensive care unit.

Mundada SD, Gosavi KS, Upasani C.

[PMID: 24700915](#) [PubMed] [PMCID: PMC3968669](#) Free PMC Article

13. Disabil Health J. 2014 Apr;7(2):251-5. doi: 10.1016/j.dhjo.2013.06.005. Epub 2013 Aug 20.

Work participation among middle-aged persons with cerebral palsy or spina bifida - a longitudinal study.

Törnbohm M1, Jonsson U2, Sunnerhagen KS3.

BACKGROUND: Most studies of work participation among persons with cerebral palsy (CP) or spina bifida (SB) have focused on young adults, little is known about older adults. **OBJECTIVE:** The aim of this study was to compare work participation in 2009 with 1997 (98). **METHODS:** Two groups of persons with CP or SB in Gothenburg, Sweden with an IQ above 70 were interviewed using a structured questionnaire regarding work participation. Group (A) was studied in 1983 (n = 55), in 1997 (n = 42) and in 2009 (n = 28). Group (B) was studied in 1998 (n = 30) and in 2009 (n = 25). In this study, the persons interviewed in 2009 were compared with their own data from 1997 (8), with a non-parametric test. **RESULTS:** Work participation had significantly decreased ($p < 0.004$) since 1997 (8); more persons worked part time or had stopped working. Thirty-eight percent had continued their education during 1997 (8)-2009, most of them worked. Of 34 persons working in 2009, 56% had wage subsidies, an increase from 42% in 1997 (8). Of the persons who worked and had continued their education, 37.5% had wage subsidies while, among persons without continued education, 72% had this support. Transportation to work functioned but not as well as in 1997 (8). More persons used transportation for people with a disability in 2009 than in 1997 (8) and criticism was expressed about the transportation system. **CONCLUSIONS:** Results showed that work participation for middle-aged persons with CP or SB without intellectual disability decreased with age but continued education and wage subsidies facilitated work participation.

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[PMID: 24680055](#) [PubMed - in process]

14. Neuropediatrics. 2014 Apr 3. [Epub ahead of print]

German Translation of the Caregiver Priorities and Child Health Index of Life with Disabilities Questionnaire: Test-Retest Reliability and Correlation with Gross Motor Function in Children with Cerebral Palsy.

Jung NH1, Brix O2, Bernius P3, Schroeder AS2, Kluger GJ4, Beyerlein A5, Weir S6, von Kries R7, Narayanan UG6, Mall V1, Berweck S2.

We aimed to translate the Caregiver Priorities and Child Health Index of Life with Disabilities (CPCHILD) questionnaire into German and to evaluate its reliability and validity by studying the association between CPCHILD scores and gross motor function as measured by the gross motor function classification system (GMFCS) in children with cerebral palsy (CP). The original CPCHILD questionnaire and manual were translated and back translated. It was administered to primary caregivers of persons with CP (GMFCS levels III-V) and was completed a second time 2 weeks after the first to measure test-retest reliability (n = 17). Primary caregivers of 68 children with CP; GMFCS level III (n = 14), level IV (n = 28), and level V (n = 26) completed the questionnaire. Mean total CPCHILD scores across GMFCS levels were 67.1 ± 14.9 for GMFCS level III, 56.6 ± 11.8 for level IV, and 44.3 ± 12.9 for level V. Good correlation ($r = -0.56$) was observed between GMFCS and total scores test-retest reliability showed intraclass correlation coefficients between 0.4 and 0.9. The German CPCHILD yielded similar test-retest reliability and score distributions across the GMFCS level as the original version. The best correlations were observed for domains that are close to the functional deficits.

Georg Thieme Verlag KG Stuttgart · New York.

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15. Res Dev Disabil. 2014 Mar 27. pii: S0891-4222(14)00109-7. doi: 10.1016/j.ridd.2014.03.017. [Epub ahead of print]

The link between impaired theory of mind and executive function in children with cerebral palsy.

Li X1, Wang K2, Wu J3, Hong Y3, Zhao J3, Feng X3, Xu M3, Wang M4, Ndasauka Y5, Zhang X6.

The aim of the study was to explore the relationship between theory of mind (ToM) deficits and executive function (EF) impairments in children with cerebral palsy (CP), 42 CP with children and 42 typically developing (TD) children, acting as controls, were assessed on the tasks of ToM (false belief and faux pas) and EF (inhibition, updating and shifting). Results showed that CP children had deficits both in ToM and EF tasks. The correlation analyses showed that two EF components (inhibition and updating) were strongly related to false belief and faux pas in both two groups. We also found correlation between shifting and false belief and faux pas. However, this correlation was only found in TD children and not in children with CP. These findings suggest that children with CP lag behind TD children in both ToM and EF. Further, the results reveal, interestingly, that ToM deficits in CP children might be related to their inhibition and updating impairments, but not to shifting impairments.

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Prevention and Cure

16. Obstet Gynecol Int. 2014;2014:976321. Epub 2014 Feb 20.

Autologous Cord Blood Therapy for Infantile Cerebral Palsy: From Bench to Bedside.

Jensen A.

About 17 million people worldwide live with cerebral palsy, the most common disability in childhood, with hypoxic-ischemic encephalopathy, preterm birth, and low birth weight being the most important risk factors. This review will focus on recent developments in cell therapy for infantile cerebral palsy by transplantation of autologous umbilical cord blood. There are only 4 publications available at present; however, the observations made along with experimental data in vivo and in vitro may be of utmost importance clinically, so that a review at an early developmental stage of this new therapeutic concept seems justified. Particularly, since the first published double-blind randomized placebo-controlled trial in a paradigm using allogeneic cord blood and erythropoietin to treat cerebral palsy under immunosuppression showed beneficial therapeutic effects in infantile cerebral palsy, long-held doubts about the efficacy of this new cell therapy are dispelled and a revision of therapeutic views upon an ailment, for which there is no cure at present, is warranted. Hence, this review will summarize the available information on autologous cord blood therapy for cerebral palsy and that on the relevant experimental work as far as potential mechanisms and modes of action are concerned.

[PMID: 24695413](#) [PubMed - as supplied by publisher] PMID: PMC3956288 Free PMC Article

17. J Pediatr. 2014 Mar 25. pii: S0022-3476(14)00174-7. doi: 10.1016/j.jpeds.2014.02.034. [Epub ahead of print]

Not Just Cerebral Palsy: Diastrophic Dysplasia Presenting as Spastic Quadriplegia.

Anbazhagan A, Benakappa A.

[PMID: 24679608](#) [PubMed - as supplied by publisher]

18. Am J Med Genet A. 2014 Apr 3. doi: 10.1002/ajmg.a.36514. [Epub ahead of print]**Autosomal recessive spastic tetraplegia caused by AP4M1 and AP4B1 gene mutation: Expansion of the facial and neuroimaging features.**

Tüysüz B1, Bilguvar K, Koçer N, Yalçinkaya C, Çağlayan O, Gül E, Sahin S, Comu S, Günel M.

Adaptor protein complex-4 (AP4) is a component of intracellular transportation of proteins, which is thought to have a unique role in neurons. Recently, mutations affecting all four subunits of AP4 (AP4M1, AP4E1, AP4S1, and AP4B1) have been found to cause similar autosomal recessive phenotype consisting of tetraplegic cerebral palsy and intellectual disability. The aim of this study was analyzing AP4 genes in three new families with this phenotype, and discussing their clinical findings with an emphasis on neuroimaging and facial features. Using homozygosity mapping followed by whole-exome sequencing, we identified two novel homozygous mutations in AP4M1 and a homozygous deletion in AP4B1 in three pairs of siblings. Spastic tetraplegia, microcephaly, severe intellectual disability, limited speech, and stereotypic laughter were common findings in our patients. All patients also had similar facial features consisting of coarse and hypotonic face, bitemporal narrowing, bulbous nose with broad nasal ridge, and short philtrum which were not described in patients with AP4M1 and AP4B1 mutations previously. The patients presented here and previously with AP4M1, AP4B1, and AP4E1 mutations shared brain abnormalities including asymmetrical ventriculomegaly, thin splenium of the corpus callosum, and reduced white matter volume. The patients also had hippocampal globoid formation and thin hippocampus. In conclusion, disorders due to mutations in AP4 complex have similar neurological, facial, and cranial imaging findings. Thus, these four genes encoding AP4 subunits should be screened in patients with autosomal recessive spastic tetraplegic cerebral palsy, severe intellectual disability, and stereotypic laughter, especially with the described facial and cranial MRI features.

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[PMID: 24700674](#) [PubMed - as supplied by publisher]**19. Am J Obstet Gynecol. 2014 Mar 26. pii: S0002-9378(14)00278-6. doi: 10.1016/j.ajog.2014.03.050. [Epub ahead of print]****Predictors of Two-Year Cognitive Performance after Laser Surgery for Twin-Twin Transfusion Syndrome.**

Vanderbilt DL1, Schragger SM2, Llanes A3, Hamilton A4, Seri I5, Chmait RH3.

OBJECTIVE: To determine risk factors for poor cognitive performance among children treated with in utero selective laser photocoagulation of communicating vessels for twin-twin transfusion syndrome (TTTS). **METHODS:** This was a prospectively enrolled cohort study. Cognitive performance at age 2 years (± 6 weeks) was assessed via Battelle Developmental Inventory 2nd Edition (BDI-2). Multilevel regression models evaluated risk factors for poor cognitive performance at shared ("pregnancy") and individual ("child") levels. In addition to development, blindness, deafness and cerebral palsy were assessed based on physical exam. A priori power analysis determined that a sample of ≥ 100 children was required for adequate statistical power (0.80). **RESULTS:** 100 children (57 families) were evaluated. Total BDI-2 score was within normal range (mean=101.3, SD=12.2), with one child having a BDI-2 of < 70 . Individual child-level risk factors for lower BDI-2 included male sex ($\beta = -0.37$, $p < 0.01$), lower head circumference ($\beta = 0.28$, $p < 0.01$), and higher diastolic blood pressure ($\beta = -0.29$, $p < 0.01$). At the pregnancy level, lower maternal education ($\beta = 0.60$, $p < 0.001$), higher Quintero stage ($\beta = -0.36$, $p < 0.01$), and lower GA at birth ($\beta = 0.30$, $p < 0.01$) were associated with worse cognitive outcomes. Donor/recipient status, GA at surgery, fetal growth restriction, and co-twin fetal demise were not risk factors. The rate of neurodevelopmental impairment (blindness, deafness, cerebral palsy, and/or a BDI-2 score < 70) was 4%. **CONCLUSION:** Overall cognitive performance quotients were in the normal range, with risk factors for poor outcomes seen at the pregnancy and child levels. Clinical and socio-economic characteristics can identify at-risk children needing additional interventions.

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[PMID: 24681290](#) [PubMed - as supplied by publisher]

20. Dev Med Child Neurol. 2014 Apr 1. doi: 10.1111/dmcn.12465. [Epub ahead of print]**Providing a population-based perspective on the perinatal factors associated with cerebral palsy.**

Kirby RS.

[PMID: 24689749](#) [PubMed - as supplied by publisher]**21. Eur J Endocrinol. 2014 Mar 31. [Epub ahead of print]****Clinical characteristics and phenotype-genotype analysis in Turkish patients with congenital hyperinsulinism; predominance of recessive KATP channel mutations.**

Demirbilek H1, Arya VB, Ozbek MN, Akinci A, Dogan M, Demirel F, Houghton J, Kaba S, Guzel F, Baran RT, Unal S, Tekkes S, Flanagan SE, Ellard S, Hussain K.

OBJECTIVE: Congenital hyperinsulinism (CHI) is the most common cause of hyperinsulinaemic hypoglycaemia in the neonatal, infancy and childhood periods. Its clinical presentation, histology and underlying molecular biology is extremely heterogeneous. The aim of this study was to describe the clinical characteristics, analyze the genotype-phenotype correlations and describe the treatment outcome of Turkish CHI patients. **DESIGN AND METHODS:** Thirty-five patients with CHI were retrospectively recruited from four large paediatric endocrine centres in Turkey. Detailed clinical, biochemical and genotype information was collected. **RESULTS:** Diazoxide-unresponsiveness was observed in nearly-half of the patients (n=17; 48.5%). Among diazoxide-unresponsive patients, mutations in ABCC8/KCNJ11 were identified in 16 (94%). Among diazoxide-responsive patients (n=18), mutations were identified in 2 patients (11%). Genotype-phenotype correlation revealed that mutations in ABCC8/KCNJ11 were associated with increased birth weight and early age of presentation. Five patients had p.L1171fs (c.3512del) ABCC8 mutations, suggestive of a founder effect. The rate of detection of a pathogenic mutation was higher in consanguineous families compared to non-consanguineous families (87.5% vs. 21%; p<0.0001). Among diazoxide-unresponsive group, 10 patients were medically managed with octreotide therapy and carbohydrate rich feeds and 6 patients underwent subtotal pancreatectomy. there was high incidence of developmental delay and cerebral palsy among diazoxide-unresponsive patients. **Conclusions:** This is the largest study to report genotype-phenotype correlations among Turkish patients with CHI. Mutations in ABCC8 and KCNJ11 are the most common causes of CHI in Turkish patients (48.6%). There is higher likelihood of genetic diagnosis in patients with early age of presentation, higher birth weight and from consanguineous pedigrees.

[PMID: 24686051](#) [PubMed - as supplied by publisher]**22. Glia. 2014 Mar 31. doi: 10.1002/glia.22658. [Epub ahead of print]****Pathophysiology of glia in perinatal white matter injury.**

Back SA1, Rosenberg PA.

Injury to the preterm brain has a particular predilection for cerebral white matter. White matter injury (WMI) is the most common cause of brain injury in preterm infants and a major cause of chronic neurological morbidity including cerebral palsy. Factors that predispose to WMI include cerebral oxygenation disturbances and maternal-fetal infection. During the acute phase of WMI, pronounced oxidative damage occurs that targets late oligodendrocyte progenitors (pre-OLs). The developmental predilection for WMI to occur during prematurity appears to be related to both the timing of appearance and regional distribution of susceptible pre-OLs that are vulnerable to a variety of chemical mediators including reactive oxygen species, glutamate, cytokines, and adenosine. During the chronic phase of WMI, the white matter displays aberrant regeneration and repair responses. Early OL progenitors respond to WMI with a rapid robust proliferative response that results in a several fold regeneration of pre-OLs that fail to terminally differentiate along their normal developmental time course. Pre-OL maturation arrest appears to be related in part to inhibitory factors that derive from reactive astrocytes in chronic lesions. Recent high field magnetic resonance imaging (MRI) data support that three distinct forms of chronic WMI exist, each of which displays unique MRI and histopathological features. These findings suggest the possibility that therapies directed at myelin regeneration and repair could be initiated early after WMI and monitored over time. These new mechanisms of acute and chronic WMI provide access to a variety of new strategies to prevent or promote repair of WMI in

premature infants. GLIA 2014.

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23. J Inherit Metab Dis. 2014 Apr 3. [Epub ahead of print]

What is new for monoamine neurotransmitter disorders?

Marecos C1, Ng J, Kurian MA.

The monoamine neurotransmitter disorders are increasingly recognized as an expanding group of inherited neurometabolic syndromes caused by disturbances in the synthesis, transport and metabolism of the biogenic amines, including the catecholamines (dopamine, norepinephrine, and epinephrine) and serotonin. Disturbances in monoamine metabolism lead to neurological syndromes that frequently mimic other conditions, such as hypoxic ischemic encephalopathy, cerebral palsy, parkinsonism-dystonia syndromes, primary genetic dystonia and paroxysmal disorders. As a consequence, neurotransmitter disorders are frequently misdiagnosed. Early and accurate diagnosis of these neurotransmitter disorders is important, as many are highly amenable to, and some even cured by, therapeutic intervention. In this review, we highlight recent advances in the field, particularly the recent extensive characterization of known neurotransmitter disorders and identification of novel neurotransmitter disorders. We also provide an overview of current and future research in the field focused on developing novel treatment strategies.

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24. Thyroid. 2014 Mar 31. [Epub ahead of print]

Thyroid hormone supplementation in Preterm Infants born before 28 weeks' gestational age and neurodevelopmental outcome at age 36 months.

van Wassenaer A1, Ares-Segura S, Golombek S, Kok JH, Paneth N, Kase J, Lagamma E.

Background: Thyroid hormones are required for normal brain maturation, and neonatal plasma thyroid hormone concentrations are low in infants < 28 weeks gestation. It is not known whether treatment of such infants with thyroid hormone improves neurodevelopmental outcome. Methods: At three years corrected age, mental, motor, and neurological development was assessed in infants born <28 weeks gestational age who had participated in a phase I trial of differing doses and modes of administration of thyroid hormone. The trial's endpoints were thyroid hormone and TSH plasma concentrations plasma in eight study arms, six treated with thyroid hormone (4, 8, and 16 microgram thyroxine (T4)/kg/d, bolus or continuous), one treated with iodine only, and one treated with placebo. Follow-up at three years was not part of the original study goals. Developmental index-scores, rates of cerebral palsy (CP) and rates of adverse outcome (death or moderate to severe delay in development and/or disabling CP) were compared between the eight study groups and between groups combined by dosage level, and between infants with and without T4 supplementation. Results: Of 166 randomized infants, 32 (19%) infants died in the neonatal period. Of the 134 survivors, follow up results were available in 89 children (66%). Mental and motor development and rates of cerebral palsy did not differ in any of the comparisons made. Conclusion: In this phase 1 study, no differences in neurodevelopment were found in relation to thyroid hormone treatment. but power was insufficient to detect any but very large differences. 247 words.

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