Cerebral palsy research news

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Professor Nadia Badawi AM
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Interventions and Management

1. Grip Strength and Sarcopenia in Children With Cerebral Palsy: A Level Playing Field?

Mahmud Fazıl Aksakal, Ahmad J Abdulsalam, Murat Kara, Levent Özçakar

Pediatr Neurol. 2024 Sep 11:161:84. doi: 10.1016/j.pediatrneurol.2024.09.002. Online ahead of print.

No abstract available

PMID: 39332074

2. Group-based progressive functional, high-intensity training in adolescents and young adults with unilateral cerebral palsy - a tool to improve gross motor function, endurance and gait? - a pilot study

Michèle Widmer, Alice Minghetti, Jacqueline Romkes, Martin Keller, Ramon Gysin, Cornelia Neuhaus, Bastian Widmer, Morgan Sangeux, Elke Viehweger

Dev Neurorehabil. 2024 Oct;27(7):235-242. doi: 10.1080/17518423.2024.2398151. Epub 2024 Sep 23.

Purpose: This pilot study assessed the safety and effects of progressive functional high-intensity training in a group setting for adolescents with unilateral cerebral palsy (CP) on daily function indicators. Methods: Nine adolescents (mean age 16.9 years, GMFCS levels I-II) participated in 12 weeks of training (2x/week). Evaluations included 3D gait analysis, the 6-min walking test (6MWT), clinical exams, and the Gross Motor Function Measure-66 (GMFM-66). Results: No adverse events occurred, and GMFM-66 scores significantly improved ($p = .031$, $\Delta = 2.19\%$). Although increases in 6MWT ($p = .09$, $\Delta = 29.8$ m) performance and propulsion ratio ($p = .067$, $\Delta = 5.4\%$) for the affected leg were observed, they were not statistically significant. Discussion: The study suggests that this training is safe and may enhance gross motor function, endurance, and gait asymmetry in unilateral CP. Future research should include upper limb evaluations and out-of-clinic motion analysis with wearable inertial measurement units (IMUs) to provide a more comprehensive assessment of functional movements.

PMID: 39311681

3. Critically appraised paper: In young children with cerebral palsy, intensive child-initiated mobility training with variability and error is as effective as conventional therapist-directed training for improving gross motor skills. [commentary]

Hércules Ribeiro Leite

J Physiother. 2024 Sep 25:S1836-9553(24)00076-6. doi: 10.1016/j.jphys.2024.08.001. Online ahead of print.

No abstract available

PMID: 39327175

4. Critically appraised paper: In young children with cerebral palsy, intensive child-initiated mobility training with variability and error is as effective as conventional therapist-directed training for improving gross motor skills [synopsis]

Nikki Milne

J Physiother. 2024 Sep 25:S1836-9553(24)00077-8. doi: 10.1016/j.jphys.2024.07.007. Online ahead of print.

No abstract available

PMID: 39327169

5. The Effect of Mobility Assistance Dogs on Quality of Life in Children with Physical and Neurological Impairments

Heather Curtin, Ciaran K Simms, Damien Kiernan, Richard B Reilly, Michelle Spirtos

Phys Occup Ther Pediatr. 2024 Sep 23:1-14. doi: 10.1080/01942638.2024.2400344. Online ahead of print.

Aims: To measure the quality of life in children with impaired walking who receive a mobility assistance dog (MAD). Methods: The parents of ten children who received a MAD completed the cerebral palsy quality of life questionnaire, before receiving their dog and at one, three, and six-month follow-up. Data were analyzed to assess changes for each participant and to the group. Results: The group showed a positive change in the domains of social well-being and acceptance, feelings about functioning, and emotional well-being and self-esteem after six months. Children with less impairment (GMFCS I-II) showed a change in social-wellbeing and acceptance, feelings about functioning, participation, physical health, and emotional-wellbeing and self-esteem after six months. Children with more impairment (GMFCS III-IV) showed no change at any timepoint measured. Conclusions: This novel therapeutic area of receiving a MAD demonstrated some positive quality of life changes after six months for a small group of children with impaired walking. These are preliminary findings in a small sample and this intervention would benefit from further study.

PMID: 39313998

6. Natural history of cerebral visual impairment in children with cerebral palsy

Jessica Galli, Erika Loi, Stefano Calza, Serena Micheletti, Anna Molinaro, Alessandra Franzoni, Andrea Rossi, Francesco Semeraro, Lotfi B Merabet, Elisa Fazzi

Dev Med Child Neurol. 2024 Sep 24. doi: 10.1111/dmcn.16096. Online ahead of print.

Aim: To longitudinally evaluate the natural history of cerebral visual impairment (CVI) in children with cerebral palsy (CP) and identify which early visual signs or symptoms are associated with cognitive visual disorders (CVDs) at school age. Method: Fifty-one individuals with CP and CVI underwent an ophthalmological, oculomotor, and basic visual function evaluation at three time points: T0 (6-35 months old); T1 (3-5 years old); and T2 (≥6 years old). We also performed a cognitive visual evaluation at T2. Logistic regression fitted using a generalized estimation equation (binary) and cumulative link models (ordinal) were used to model the outcomes of interest. Results: Ophthalmological deficits were stable over time, except for ocular fundus abnormalities (T1-T0, $p = 0.01$; T2-T1, $p = 0.02$; T2-T0, $p < 0.01$) and strabismus, whose frequency increased with age (T2-T0, $p= 0.02$ with T2-T0, $p = 0.05$). Conversely, fixation (T1-T0, T2-T0, $p < 0.01$), smooth pursuit (T2-T1, T2-T0, $p < 0.01$), saccades (T1-T0, T2-T1, T2-T0, $p < 0.01$), as well as visual acuity, contrast sensitivity, and visual field (T1-T0, T2-T0, $p < 0.01$) all improved over time. Early oculomotor dysfunction was associated with CVD at T2. Interpretation: Although a diagnosis of CVI was confirmed in all children at each time point, several visual signs and symptoms improved over time; in some cases, they reached complete recovery at T1 and T2. These results emphasize the 'permanent' but 'not unchanging' nature of the CVI associated with CP during development.

PMID: 39316724

7. Rib-on-pelvis deformity: a modifiable driver of pain and poor health-related quality of life in cerebral palsy

Vineet M Desai, Margaret Bowen, Jason B Anari, John M Flynn, Burt Yaszay, Paul Sponseller, Mark Abel, Joshua Pahys; Harms Study Group; Patrick J Cahill

Spine Deform. 2024 Sep 26. doi: 10.1007/s43390-024-00974-0. Online ahead of print.

Purpose: Cerebral Palsy (CP) often presents with a sweeping thoracolumbar scoliosis and pelvic obliquity. With severe pelvic obliquity, the ribs come into contact with the high side of the oblique pelvis, termed rib-on-pelvis deformity (ROP). ROP can result in costo-iliac impingement, or pain associated with ROP, and can also adversely affect breathing and sitting balance. The

goal of this study was to evaluate whether CP patients with ROP have worse health-related quality of life (HRQOL) before surgery and a greater improvement in HRQOL after surgery. Methods: A retrospective analysis of a prospectively collected, multicenter, international registry was performed for all nonambulatory patients with CP treated with spinal fusion with at least two-year follow-up. HRQOL was measured via the Caregiver Priorities & Child Health Index of Life with Disabilities (CPCHILD) questionnaire domains (0 = most disability, 100 = least disability). ROP was defined as having a rib distal to the superior portion of the iliac crest on preop upright radiographs. The ROP group and control group without ROP were compared regarding six domain scores and total score of CPCHILD. Multiple linear regression was used to control for curve apex location, major coronal Cobb angle, type of tone, and pelvic obliquity. Results: 340 patients met inclusion criteria (52% female, mean age 14.0 years). The mean major coronal Cobb angle was 81 degrees and mean pelvic obliquity was 22 degrees. 176 patients (51.8%) had ROP while 164 patients (48.2%) did not. ROP was independently associated with worse preoperative Positioning/Transfers/Mobility (PTM), Comfort & Emotions (C&E), and total CPCHILD score via the CPCHILD questionnaire ($p < 0.05$). Patients with preoperative ROP experienced a greater improvement in the C&E and PTM domains as well as total CPCHILD score than patients without ROP ($p < 0.05$). Conclusion: CP patients with rib-on-pelvis deformity experience more pain and worse HRQOL than patients without this deformity. These patients experienced a greater improvement in HRQOL after spinal fusion measured via the CPCHILD questionnaire.

PMID: 39325331

8. [Sleep in chronic neuropediatric diseases] [Article in Spanish] [Abstract in English, Spanish]

Alberto Navarro Vergara, Oscar Sans Capdevilla, Gabriel González Rabelino

Review Medicina (B Aires). 2024 Sep:84 Suppl 3:93-98.

The prevalence of sleep disorders (SD) is notoriously increased in children with chronic neurological disease, with a negative bidirectional link that aggravates their symptomatology and has a negative impact on the quality of life of the child and their families. Identifying and recognizing this association is key for the child neurologist since the treatment of SD significantly improves daytime symptomatology in neurodevelopmental disorders, epilepsy, primary headaches, cerebral palsy and neuromuscular diseases.

PMID: 39331783

9. Negative emotions reduce sensorimotor cortex activity during proprioceptive modulation of rolandic ~20HZ beta rhythm in typically developing children and those with neurodevelopmental conditions

Álvaro Sabater-Gárriz, Francesc Mestre-Sansó, Vicent Canals, José Antonio Mingorance, Pedro Montoya, Inmaculada Riquelme

Res Dev Disabil. 2024 Sep 25:154:104842. doi: 10.1016/j.ridd.2024.104842. Online ahead of print.

Background: The Rolandic ~20-Hz beta rhythm of the sensorimotor cortex is associated with motor function and perception. However, the modulation of this rhythm by different emotional stimuli is an innovative area of research. Aims: This study aims at investigating the impact of affective pictures (positive, negative, and neutral) on the proprioceptive modulation of the Rolandic ~20 Hz beta rhythm in typically developing children and children with neurodevelopmental disorders (i.e. cerebral palsy and autism). Methods and procedures: EEG was recorded while participants experienced passive wrist movements during the simultaneous viewing of affective pictures. Time-frequency analysis of the sensorimotor oscillatory activity was performed. Outcomes and results: Our findings revealed that pictures with negative emotional valence notably diminish event-related synchronization (ERS) amplitude during the perception of hand movement in all groups of children. Conclusions and implications: These findings suggest that emotional stimuli, particularly the negative ones, could significantly influence brain's processing of proprioception, adding knowledge to the interaction of common comorbidities, such as sensorimotor disorders and emotional dysregulation, in children with neurodevelopmental disabilities.

PMID: 39326290

10. Validation of two novel human activity recognition models for typically developing children and children with Cerebral Palsy

Marte Fossflaten Tørring, Aleksej Logacjov, Siri Merete Brændvik, Astrid Ustad, Karin Roeleveld, Ellen Marie Bardal

PLoS One. 2024 Sep 23;19(9):e0308853. doi: 10.1371/journal.pone.0308853. eCollection 2024.

Human Activity Recognition models have potential to contribute to valuable and detailed knowledge of habitual physical activity for typically developing children and children with Cerebral Palsy. The main objective of the present study was to develop and validate two Human Activity Recognition models. One trained on data from typically developing children (n =

63), the second also including data from children with Cerebral Palsy (n = 16), engaging in standardised activities and free play. Our data was collected using accelerometers and ground truth was established with video annotations. Additionally, we aimed to investigate the influence of window settings on model performance. Utilizing the Extreme gradient boost (XGBoost) classifier, twelve sub-models were created, with 1-,3- and 5-seconds windows, with and without overlap. Both Human Activity Recognition models demonstrated excellent predictive capabilities (>92%) for standardised activities for both typically developing and Cerebral Palsy. From all window sizes, the 1-second window performed best for all test groups. Accuracy was slightly lower (>75%) for the Cerebral Palsy test group performing free play activities. The impact of window size and overlap varied depending on activity. In summary both Human Activity Recognition models effectively predict standardised activities, surpassing prior models for typically developing and children with Cerebral Palsy. Notably, the model trained on combined typically developing children and Cerebral Palsy data performed exemplary across all test groups. Researchers should select window settings aligned with their specific research objectives.

PMID: 39312531

11. Biomechanical Gait Analysis Using a Smartphone-Based Motion Capture System (OpenCap) in Patients with Neurological Disorders

Yu-Sun Min, Tae-Du Jung, Yang-Soo Lee, Yonghan Kwon, Hyung Joon Kim, Hee Chan Kim, Jung Chan Lee, Eunhee Park

Bioengineering (Basel). 2024 Sep 12;11(9):911. doi: 10.3390/bioengineering11090911.

This study evaluates the utility of OpenCap (v0.3), a smartphone-based motion capture system, for performing gait analysis in patients with neurological disorders. We compared kinematic and kinetic gait parameters between 10 healthy controls and 10 patients with neurological conditions, including stroke, Parkinson's disease, and cerebral palsy. OpenCap captured 3D movement dynamics using two smartphones, with data processed through musculoskeletal modeling. The key findings indicate that the patient group exhibited significantly slower gait speeds (0.67 m/s vs. 1.10 m/s, p = 0.002), shorter stride lengths (0.81 m vs. 1.29 m, p = 0.001), and greater step length asymmetry (107.43% vs. 91.23%, p = 0.023) compared to the controls. Joint kinematic analysis revealed increased variability in pelvic tilt, hip flexion, knee extension, and ankle dorsiflexion throughout the gait cycle in patients, indicating impaired motor control and compensatory strategies. These results indicate that OpenCap can effectively identify significant gait differences, which may serve as valuable biomarkers for neurological disorders, thereby enhancing its utility in clinical settings where traditional motion capture systems are impractical. OpenCap has the potential to improve access to biomechanical assessments, thereby enabling better monitoring of gait abnormalities and informing therapeutic interventions for individuals with neurological disorders.

PMID: 39329653

12. Compare the effectiveness of two treatments, behavioral activation and acceptance and commitment therapy, on depression and mental rumination in mothers of children with cerebral palsy in Ilam city

Mostafa Alirahmi, Sattar Kikhavani, Homeira Soleimannejad

J Educ Health Promot. 2024 Jul 29:13:260. doi: 10.4103/jehp.jehp_1720_23. eCollection 2024.

Background: Since in most families, mothers are more responsible for taking care of children and have more responsibility than fathers for monitoring the child, taking care of a disabled child can have a more negative effect on the psychological state of mothers. This study aimed to investigate the effectiveness of behavioral activation (BA) and acceptance and commitment therapy (ACT) in depression and rumination in mothers with children with cerebral palsy. Materials and methods: This research was quasi-experimental field research with a pre-posttest and control group. The study population comprised 237 mothers with children who had cerebral palsy and were referred to occupational therapy centers in Ilam, Iran. The sample consisted of 60 participants selected by convenience sampling, who were randomly divided into two experimental groups (BA and ACT) and a control group (n = 20 per group). Data were analyzed using Statistical Package for the Social Sciences (SPSS) version 27.0 with descriptive and inferential statistics, such as mean, standard deviation, and multivariate analysis of covariance (MANCOVA). Results: The results of both BA and ACT were effective in reducing depression ($P < 0.01$) and rumination ($P < 0.01$) in the two experimental groups compared with the control group in the posttest phase. Furthermore, the effectiveness of the ACT in reducing depression and rumination was found to be more significant than the effectiveness of BA ($P \leq 0.05$). Conclusion: The findings of the study suggest that both BA and ACT are effective in reducing depression and rumination in the research participants. However, the study also found that ACT was more effective than BA in reducing depression and rumination. Thus, both approaches can be used to strengthen treatment interventions to lessen depression and rumination in the research participants.

PMID: 39310014

13. Maternal Diabetes and Neurodevelopmental Outcomes of Infants Born Before 29 Weeks' Gestation

Adetokunbo Akinseye, Christy Pylypjuk, Diane Moddemann, Jehier Afifi, Rudaina Banihani, Khalid Aziz, Dianna Wang, Mary Seshia; Canadian Neonatal Network, Canadian Neonatal Follow-up Network under the umbrella of Canadian Preterm Birth Network

J Pediatr. 2024 Sep 19:114319. doi: 10.1016/j.jpeds.2024.114319. Online ahead of print.

Objective: To compare the neurodevelopmental outcomes of infants born at <29 weeks' gestation and exposed to diabetes in pregnancy with those unexposed. Study design: This was a retrospective cohort study using the Canadian Neonatal Network (CNN) and Canadian Neonatal Follow-Up Network (CNFUN) databases. Infants born <29 weeks' gestation and admitted to a level 3 NICU from 2009 through 2018 who had neurodevelopmental assessments at 18 to 24 months corrected age (CA) were eligible. The two primary outcomes were: i) Neurodevelopmental Impairment (NDI) (≥1 of Bayley-III scores < 85 in any domain, cerebral palsy, or vision or hearing impairment); and ii) significant NDI (sNDI) (≥1 of Bayley-III scores < 70 in any domain, cerebral palsy GMFCS ≥ 3, bilateral blindness, or need for hearing aids or cochlear implants). Secondary outcomes were the individual components of NDI and sNDI. Adjusted odds ratios with 95% CIs were calculated to determine outcomes between groups. Results: Of 13,988 eligible infants, 55% attended neurodevelopmental follow-up assessments. Infants exposed to diabetes had increased odds of NDI compared with those unexposed (aOR 1.09 (95% CI 1.08-1.54); there was no difference in sNDI (aOR 1.07 (95% CI 0.84-1.36). Language and motor delays were more common in those exposed to maternal diabetes. Conclusions: Higher rates of NDI, language, or motor delays were present in infants born at < 29 weeks' gestation exposed to diabetes in utero. Future research is needed to determine the etiology and clinical significance of these findings.

PMID: 39306321

14. Functional classification systems in Brazilian children with cerebral palsy: Reliability and associations between functional levels

Kennea Martins Almeida Ayupe, Amanda Larissa Oliveira Lima, Gabrielly Cristine de Alcântara Gomes, Deysiane Sobrinho de Sousa, Ana Cristina Resende Camargos, Paula Silva de Carvalho Chagas, Hércules Ribeiro Leite, Egmar Longo, Aline Martins de Toledo

Dev Neurorehabil. 2024 Oct;27(7):243-250. doi: 10.1080/17518423.2024.2398158. Epub 2024 Sep 23.

Children with Cerebral palsy (CP) present movement and posture disorders. The Gross Motor Function Classification System (GMFCS), Manual Ability Classification System (MACS), Communication Function Classification System (CFCS), Eating and Drinking Ability Classification System (EDACS), and Visual Function Classification System (VFCS) enhance the understanding of their performance. We verified inter-rater reliability and associations between the classification levels. Physical therapists classified 100 Brazilian children with CP (3-17 years) according to GMFCS, MACS, CFCS, EDACS, and VFCS. To evaluate inter-rater reliability (Intraclass Correlation Coefficient-ICC) two independent examiners concurrently assessed a subset of 60 participants. According to Spearman's correlation coefficients, there were associations between GMFCS/MACS (r = 0.81), GMFCS/CFCS (r = 0.70), MACS/CFCS (r = 0.73), GMFCS/VFCS (r = 0.61), MACS/VFCS (r = 0.61), CFCS/EDACS (r = 0.58), CFCS/VFCS (r = 0.50), and EDACS/VFCS (r = 0.45) ($p < .05$). The inter-rater reliability ranged from excellent (ICC = 0.93-0.99) to good (ICC = 0.89), $p < .05$. The classification systems are reliable, and the levels associated with each other in Brazilian children, especially the GMFCS, MACS, and CFCS.

PMID: 39311709

15. Multi-center improvement in screening for dystonia in young people with cerebral palsy

Bhooma R Aravamuthan, Emma Lott, Esra Pehlivan, Keerthana Chintalapati, Deborah Grenard, Desiree Roge, Rose Gelineau-Morel, Dante Kyle, Christie Becu, Michael Kruer, Linn Katus, Paul Gross, Amy Bailes; Cerebral Palsy Research Network

medRxiv [Preprint]. 2024 Sep 14:2024.09.13.24313431. doi: 10.1101/2024.09.13.24313431.

Background and objectives: Dystonia is a common, debilitating, and often treatment refractory motor symptom of cerebral palsy (CP), affecting 70-80% of this population based on research assessments. However, routine clinical evaluation for dystonia in CP has failed to match these expected numbers. Addressing this diagnostic gap is a medical imperative because the presence of dystonia rules in or out certain treatments for motor symptoms in CP. Therefore, our objective was to optimize rates of clinical dystonia screening to improve rates of clinical dystonia diagnosis. Methods: Using the quality improvement (QI) infrastructure of the Cerebral Palsy Research Network (CPRN), we developed and implemented interventions to increase the documentation percentage of five features of dystonia in young people with CP, aged 3-21 years old. This QI initiative was implemented by seven physiatry and pediatric movement disorders physicians at four tertiary-care pediatric hospitals between 10/10/21 and 7/1/23. We collected visit data cross-sectionally across all participating sites every 2 weeks and tracked our progress using control charts. Results: We assessed 847 unique visits, mostly for established patients (719/847, 85%) who were 9.2 years old on average (95% CI 8.8-9.5). By 4/10/22, the mean percentage of dystonia screening elements documented across

all sites rose from 39% to 90% and the mean percentage of visits explicitly documenting the presence or absence of dystonia rose from 65% to 94%. By 10/23/22, the percentage of visits diagnosing dystonia rose from 57% to 74%. These increases were all sustained through the end of the study period in 7/1/23. Discussion: Using a rigorous QI-driven process across four member sites of a North American learning health network (CPRN), we demonstrated that we could increase screening for dystonia and that this was associated with increased clinical dystonia diagnosis, matching expected research-based rates. We propose that similar screening should take place across all sites caring for people with CP.

PMID: 39314964

16. Motor development trajectories of children with cerebral palsy in a community-based early intervention program in rural South India

Marie Brien, Dinesh Krishna, Ramasubramanian Ponnusamy, Cathy Cameron, Rahim Moineddin, Franzina Coutinho

Res Dev Disabil. 2024 Sep 24:154:104829. doi: 10.1016/j.ridd.2024.104829. Online ahead of print.

Background: Developmental trajectories are crucial for evidence-based prognostication, planning interventions, and monitoring progress in children with cerebral palsy (CP). Aims: To describe gross motor development patterns of children with CP in rural South India for the five Gross Motor Function Classification System (GMFCS) levels. Methods: Longitudinal cohort study of 302 children (176 males, 126 females) with CP aged 0 to 10 years, followed by a community-based early intervention program. GMFCS levels were 5.4 % level 1, 16.5 % level II, 22.8 % level III, 26.8 % level IV, and 28.5 % level V. Assessments were undertaken using the Gross Motor Function Measure (GMFM-66) at 6-month intervals between April 2017 and August 2020. Longitudinal analyses were performed using mixed-effects linear regression models. Outcomes and results: Five distinct motor development curves were created for ages 0 to 10 years by GMFCS levels as a function of age and GMFM-66 with a stable limit model, variation in estimated limits and rates of development. Conclusions and implications: Motor development trajectories for CP in an LMIC differ from those reported in HICs. Consideration of how social determinants of health, environmental and personal factors impact motor development in low-resource contexts is crucial. Further work is needed to describe developmental trajectories of children for CP in LMICs.

PMID: 39321692

17. Proxy- and self-report evaluation of quality of life in cerebral palsy: Using Spanish version of CPQOL for Children and adolescents

Alba Aza, Inmaculada Riquelme, María Gómez Vela, Marta Badia

Res Dev Disabil. 2024 Sep 24:154:104844. doi: 10.1016/j.ridd.2024.104844. Online ahead of print.

Background: Promoting quality of life (QoL) is one of the main goals in interventions carried out with children and adolescents with cerebral palsy (CP). Aims: The aim of this study was to analyze the determinants of QoL in children with CP, including evaluations by the children themselves and their parents, and to identify discrepancies between evaluators. Methods and procedures: The adapted Spanish version of the Cerebral Palsy Quality of Life (CP-QOL) for children and adolescents (self-report and primary caregiver-reports versions) was applied to a sample of 74 children with CP and their respective parents (totaling 222 participants), as well as instruments to measure functioning (i.e., GMFCS, MACS, CFCS and EDACS). The average age of the children was 12.50 (SD=4.07), with a higher number of boys (55.7 %). Outcomes and results: The lowest QoL levels were found in the Functional dimension in both assessments (Mchildren/adolescents=70.21, Mparents=58.14). For children, the highest rated dimension was Social Well-being (M=74.54), while for parents it was School (M=71.03). The degree of agreement between evaluators was low in almost all dimensions (ICC≤.40). More satisfactory predictive models were constructed from the evaluations carried out by parents, except in the case of the Access to Services dimension, with functioning measures being the main predictors of QoL levels. Conclusions and implications: The CP-QOL in its two available versions is a useful and specific instrument for assessing QoL in children with CP in both research and professional fields.

PMID: 39321691

18. Selective dorsal rhizotomy after baclofen intrathecal pump removal: a single-center experience and review of the literature

Claudio Ruggiero, Massimiliano Porzio, Francesco Tengattini, Giuseppe Cinalli, Pietro Spennato

Childs Nerv Syst. 2024 Sep 24. doi: 10.1007/s00381-024-06618-y. Online ahead of print.

Purpose: Selective dorsal rhizotomy (SDR) and intrathecal baclofen (ITB) pump placement are two surgical options in children affected by spasticity secondary to cerebral palsy 1. The latest literature is an enlarging indication for SDR in case of ITB failure in non-ambulant patients as an alternative to pump reimplantation to reduce spasticity and facilitate patients' care.

Methods: A retrospective single-center study has screened all children diagnosed with spastic tetraparesis who underwent in the last 10 years SDR and had previously ITB pump implanted. A cohort of six patients was pooled out. Furthermore, pertinent literature has been reviewed. Results: Indication for pump removal was pump pocket infection, parents' decision, and poor response to ITB. Patients' amount of lifetime with the pump implanted has been 6.9 years. The preoperative and postoperative Ashworth scores were statistically different in both procedures (p = 0.005 and p = 0.,02). Conclusions: Only two studies investigated the pediatric population undergoing SDR in the occurrence of ITB pump removal. Authors are offering SDR to all children, regardless of GMFCS groups and previous ITB pump placement. In conclusion, SDR represents a valid tool in neurosurgeon's hands to help ameliorate patients' long-lasting quality of life, reducing the severity of the spasticity and leading to better management by caregivers.

PMID: 39317766

19. EEG-based sensorimotor neurofeedback for motor neurorehabilitation in children and adults: A scoping review

Elena Cioffi, Anna Hutber, Rob Molloy, Sarah Murden, Aaron Yurkewich, Adam Kirton, Jean-Pierre Lin, Hortensia Gimeno, Verity M McClelland

Clin Neurophysiol. 2024 Aug 20:167:143-166. doi: 10.1016/j.clinph.2024.08.009. Online ahead of print.

Objective: Therapeutic interventions for children and young people with dystonia and dystonic/dyskinetic cerebral palsy are limited. EEG-based neurofeedback is emerging as a neurorehabilitation tool. This scoping review maps research investigating EEG-based sensorimotor neurofeedback in adults and children with neurological motor impairments, including augmentative strategies. Methods: MEDLINE, CINAHL and Web of Science databases were searched up to 2023 for relevant studies. Study selection and data extraction were conducted independently by at least two reviewers. Results: Of 4380 identified studies, 133 were included, only three enrolling children. The most common diagnosis was adult-onset stroke (77%). Paradigms mostly involved upper limb motor imagery or motor attempt. Common neurofeedback modes included visual, haptic and/or electrical stimulation. EEG parameters varied widely and were often incompletely described. Two studies applied augmentative strategies. Outcome measures varied widely and included classification accuracy of the Brain-Computer Interface, degree of enhancement of mu rhythm modulation or other neurophysiological parameters, and clinical/motor outcome scores. Few studies investigated whether functional outcomes related specifically to the EEG-based neurofeedback. Conclusions: There is limited evidence exploring EEG-based sensorimotor neurofeedback in individuals with movement disorders, especially in children. Further clarity of neurophysiological parameters is required to develop optimal paradigms for evaluating sensorimotor neurofeedback. Significance: The expanding field of sensorimotor neurofeedback offers exciting potential as a non-invasive therapy. However, this needs to be balanced by robust study design and detailed methodological reporting to ensure reproducibility and validation that clinical improvements relate to induced neurophysiological changes.

PMID: 39321571

20. Central Nervous System Fungal Diseases in Children with Malignancies: A 16-Year Study from the Infection Working Group of the Hellenic Society of Pediatric Hematology Oncology

Loizos Petrikkos, Maria Kourti, Kondylia Antoniadi, Tatiana-Sultana Tziola, Angeliki-Eleni Sfetsiori, Vasiliki Antari, Sofia Savoukidou, Georgia Avgerinou, Maria Filippidou, Eugenia Papakonstantinou, Sophia Polychronopoulou, Emmanuel Hatzipantelis, Dimitrios Doganis, Antonios Kattamis, Vassilios Papadakis, Emmanuel Roilides, Athanasios Tragiannidis

J Fungi (Basel). 2024 Sep 14;10(9):654. doi: 10.3390/jof10090654.

We analyzed data on pediatric invasive fungal diseases of the central nervous system (CNS-IFDs) reported by five of a total of eight Pediatric Hematology-Oncology Departments in Greece for 16 years (2007-2022). A total of twelve patients (11 boys, median age: 9.5 years, range: 2-16) were reported suffering from CNS-IFDs. The underlying malignancy was acute lymphoblastic leukemia in 9/12 and acute myeloid leukemia, Ewing sarcoma, and rhabdomyosarcoma in one each. Eleven patients presented with CNS-related symptoms (i.e., seizures, headache, cerebral palsy, ataxia, hallucination, seizures, blurred vision, amaurosis). All patients had pathological MRI findings. Multifocal fungal disease was observed in 6/12 patients. Nine proven and three probable CNS-IFD cases were diagnosed. Causative pathogens in proven cases were Aspergillus spp. and Candida albicans (n = 2 each), Mucor spp., Rhizopus arrhizus, Absidia spp., Fusarium oxysporum and Cryptococcus neoformans (n = 1 each). Causative pathogens in probable cases were Aspergillus spp. (n = 2) and Candida spp. (n = 1). All patients received appropriate antifungal therapy (median duration: 69.5 days, range 19-364). Two patients underwent additional surgical treatment. Six patients were admitted to the Intensive Care Unit due to complications. Three patients (25%) died, two due to IFD and one due to an underlying disease. Early recognition and prompt intervention of CNS-IFDs may rescue the patients and improve overall survival.

PMID: 39330414

21. Case Report of Friedreich's Ataxia and ALG1-Related Biochemical Abnormalities in a Patient With Progressive Spastic Paraplegia

Aisling Quinlan, Lance Rodan, Elizabeth Barkoudah, Amy Tam, Afshin Saffari, Ibrahim Shammas, Wasantha Ranatunga, Eva Morava-Kozicz, Devin Oglesbee, Gerald Berry, Darius Ebrahimi-Fakhari, Siddharth Srivastava

Case Reports Am J Med Genet A. 2024 Sep 26:e63890. doi: 10.1002/ajmg.a.63890. Online ahead of print.

Frataxin is an evolutionarily conserved mitochondrial protein responsible for iron homeostasis and metabolism. A deficiency of frataxin (encoded by FXN) leads to Friedreich's ataxia (FRDA), a progressive disorder that affects both the central and peripheral nervous systems, most commonly via a pathogenic GAA trinucleotide expansion. In contrast, pathogenic variants in ALG1 in humans cause a form of congenital disorder of glycosylation. Here, we present a 15-year-old boy with a clinical presentation that raised concern for complex hereditary spastic paraplegia (HSP), with motor features including progressive spastic paraparesis, cervical dystonia, cerebellar dysfunction, and diminished lower extremity reflexes. The proband was initially found to have a novel compound heterozygous variant in ALG1 on exome sequencing, along with N-glycan profiling revealing evidence of defective mannosylation and Western blot analysis demonstrating an 84% reduction in ALG1 expression. Although several of his clinical features could be explained by the ALG1 variant specifically or considered as part of the presentation of CDGs in general, there were additional phenotypes that suggested an alternative, or additional, genetic diagnosis. Subsequently, he was found to have biallelic pathogenic GAA repeat expansions in FXN on genome sequencing, leading to a diagnosis of FRDA. Given that FRDA explained all his clinical features, the ALG1 variant may have been a hypomorphic form and/or a biochemical phenotype. Our findings underscore the importance of considering FRDA as a differential diagnosis in cases of complex HSP and demonstrate the utility of unbiased genome sequencing approaches that include detection of trinucleotide repeat expansions for progressive motor disorders.

PMID: 39324476

22. Survey of the Landscape of Society Practice Guidelines for Genetic Testing of Neurodevelopmental Disorders

Siddharth Srivastava, Jordan J Cole, Julie S Cohen, Maya Chopra, Hadley Stevens Smith, Matthew A Deardorff, Ernest Pedapati, Brian Corner, Julia S Anixt, Shafali Jeste, Mustafa Sahin, Christina A Gurnett, Colleen A Campbell; Intellectual and Developmental Disabilities Research Center (IDDRC) Workgroup on Advocating for Access to Genomic Testing

Review Ann Neurol. 2024 Sep 25. doi: 10.1002/ana.27045. Online ahead of print.

Genetic testing of patients with neurodevelopmental disabilities (NDDs) is critical for diagnosis, medical management, and access to precision therapies. Because genetic testing approaches evolve rapidly, professional society practice guidelines serve an essential role in guiding clinical care; however, several challenges exist regarding the creation and equitable implementation of these guidelines. In this scoping review, we assessed the current state of United States professional societies' guidelines pertaining to genetic testing for unexplained global developmental delay, intellectual disability, autism spectrum disorder, and cerebral palsy. We describe several identified shortcomings and argue the need for a unified, frequently updated, and easily-accessible cross-specialty society guideline. ANN NEUROL 2024.

PMID: 39319594

23. In-Home Intravenous Dexmedetomidine Infusion for Treatment of Dystonia: A Pediatric Case Report

Gwendolyn J Richner, Stephanie S Allen, Lindsay M Adair, Daniel H Grossoehme, Catherine A Kelly-Langen, Wendy J McNair, Madelyne L Williams, Sarah Friebert

Case Reports A A Pract. 2024 Sep 23;18(9):e01851. doi: 10.1213/XAA.0000000000001851. eCollection 2024 Sep 1.

The off-label use of an intravenous dexmedetomidine infusion is an option for refractory symptoms in pediatric palliative care with a few published cases of in-home use, most limited to end-of-life care. After an intrathecal baclofen pump malfunction with meningitis and medication withdrawal, a 17-year-old adolescent with quadriparetic cerebral palsy and paroxysmal autonomic instability experienced relief of refractory pain and dystonia with an in-home intravenous dexmedetomidine infusion. His interdisciplinary care team collaborated to establish safety measures and adjust his home medication regimen to further improve quality of life well before his end of life.

PMID: 39311454

24. COL4A1 Gene Mutation Masquerading as Cerebral Palsy: Report of a Rare Case

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Case Reports Cureus. 2024 Aug 20;16(8):e67351. doi: 10.7759/cureus.67351. eCollection 2024 Aug.

The Collagen Type 4 alpha 1 (COL4A1), is an important component of nearly all vascular basement membranes. Pathogenic mutation of this gene results in varied manifestations. In this report, we describe a two-and-a-half-year-old boy with an eventful perinatal period, global developmental delay, and epileptic spasms. Examination revealed microcephaly, nystagmus, and spasticity in limbs. Electroencephalogram showed multifocal epileptiform discharges and MRI brain demonstrated periventricular white matter changes, intracerebral bleeds, and porencephalic cysts. CT brain showed intracranial calcifications and screening for congenital infection was negative. The molecular genetic evaluation was later confirmed with a heterozygous mutation of the COL4A1 gene on exon 37 (variant - p.Gly1050Ala) with an autosomal dominant inheritance pattern. Currently, the child has developed drug-refractory epilepsy requiring polypharmacy and the ketogenic diet. COL4A1 gene mutations are close mimickers of Cerebral Palsy, hence a high index of suspicion should be exercised while approaching a child with spastic quadriplegia in order to promptly diagnose and manage such children for a better neurological outcome.

PMID: 39310498

Prevention and Cure

25. RidStress 2 randomised controlled trial protocol: an Australian phase III clinical trial of intrapartum sildenafil citrate or placebo to reduce emergency caesarean birth for fetal distress in women with small or suboptimally grown infants at term (≥37 weeks)

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BMJ Open. 2024 Sep 25;14(9):e082945. doi: 10.1136/bmjopen-2023-082945.

Introduction: Small for gestational age (SGA) infants are at increased risk of fetal distress in labour requiring emergency operative birth (by caesarean section (CS), vacuum or forceps). We have previously shown that maternal oral sildenafil citrate (SC) in labour halves the need for operative birth for suspected fetal distress in women with appropriately grown term infants. Methods and analysis: RidStress 2 is a phase III randomised, double-blinded, placebo-controlled trial of 660 women with an SGA or suboptimally grown fetus (estimated fetal weight or abdominal circumference<10th centile for gestational age) planning a vaginal birth at term. The trial will determine whether oral intrapartum SC (50 mg eight hourly) reduces the relative risk of emergency CS for fetal distress compared with placebo. The primary outcome is CS for fetal distress, and the secondary outcomes are any operative birth for fetal distress, cost-effectiveness of SC treatment and 2-year childhood neurodevelopmental outcomes. To detect a 33% reduction in the primary outcome from 30% to 20% for an alpha of 0.05 and power of 80% with 10% dropout, requires approximately 660 women (330 in each arm). This sample size will also yield >90% power to detect a similar reduction for the secondary outcome of any operative birth (CS or instrumental vaginal birth) for fetal distress. Ethics and dissemination: Ethics approval was granted by the Mater Misericordiae Limited Human Research Ethics Committee (EC00332) on 11 September 2020. We plan to disseminate the results of this randomised controlled trial through presentations at scientific meetings and peer-reviewed journals, adhering to all relevant reporting guidelines. Trial registration number: RidStress 2 is registered with the Australian New Zealand Clinical Trials Registry (ACTRN12621000354886, 29/03/2021) and the Therapeutic Goods Association of Australia (date registered: 16 March 2021).

PMID: 39322593

26. Lung Recruitment Before Surfactant Administration in Extremely Preterm Neonates: 2-Year Follow-Up of a Randomized Clinical Trial

Francesca Gallini, Domenico Umberto De Rose, Roberta Iuliano, Domenico Marco Romeo, Milena Tana, Angela Paladini, Francesca Paola Fusco, Stefano Nobile, Francesco Cota, Chiara Tirone, Claudia Aurilia, Alessandra Lio, Alice Esposito, Simonetta Costa, Vito D'Andrea, Maria Luisa Ventura, Virgilio Carnielli, Carlo Dani, Fabio Mosca, Monica Fumagalli, Gianfranco Scarpelli, Lucio Giordano, Valeria Fasolato, Flavia Petrillo, Pasqua Betta, Agostina Solinas, Eloisa Gitto, Giancarlo Gargano, Giovanna Mescoli, Stefano Martinelli, Sandra Di Fabio, Italo Bernardo, Lucia Gabriella Tina, Alex Staffler, Ilaria Stasi, Isabella Mondello, Eleonora Scapillati, Stefania Vedovato, Gianfranco Maffei, Adriano Bove , Marcello Vitaliti, Gianluca Terrin, Paola Lago, Camilla Gizzi, Chiara Strozzi, Paolo Ernesto Villani, Alberto Berardi, Caterina Cacace, Giorgio Bracaglia, Eleonora Pascucci, Filip Cools, Jane J Pillow, Graeme Polglase, Roberta Pastorino, Anton H van Kaam, Eugenio Mercuri, Luigi Orfeo, Giovanni Vento; IN-REC-SUR-E Study Group; Silvia Malguzzi, Camilla Rigotti, Alessandra Cecchi, Gabriella Nigro, Carmine Deni Costabile, Enza Roma, Paola Sindico, Rita Venafra, Carmine Mattia, Maria Conversano, Elisa Ballardini, Alessandro Manganaro, Eleonora Balestri, Claudio Gallo, Piero Catenazzi, Maria Graziana Astori, Eugenia Maranella, Carolina Grassia, Kim Maiolo, Danilo Castellano, Luca Massenzi, Elisabetta Chiodin, Maria Rita Gallina, Chiara Consigli, Elena Sorrentino, Silvia Bonato, Monica Mancini, Roberto Perniola, Silvia Giannuzzo, Elisa Tranchina, Viviana Cardilli, Lucia Dito, Daniela Regoli, Francesca Tormena, Nadia Battajon, Roberta Arena, Benedetta Allais,

Isotta Guidotti, Federica Roversi, Valerio Meli, Viviana Tulino, Alessandra Casati

Randomized Controlled Trial JAMA Netw Open. 2024 Sep 3;7(9):e2435347. doi: 10.1001/jamanetworkopen.2024.35347.

Importance: A multicenter randomized clinical trial (RCT) showed a lung recruitment maneuver using high-frequency oscillatory ventilation just before surfactant administration (ie, intubate-recruit-surfactant-extubate [IN-REC-SUR-E]) improved the efficacy of treatment compared with the standard intubate-surfactant-extubate (IN-SUR-E) technique without increasing the risk of adverse neonatal outcomes. Objective: To examine follow-up outcomes at corrected postnatal age (cPNA) 2 years of preterm infants previously enrolled in an RCT and treated with IN-REC-SUR-E or IN-SUR-E in 35 tertiary neonatal intensive care units. Design, setting, and participants: This was a follow-up study of infants recruited into the primary RCT from 2015 to 2018 at 35 tertiary neonatal intensive care units (NICUs) in Italy. Follow-up examinations included neurodevelopmental, growth, and respiratory outcomes of these children at cPNA 2 years. Participants included spontaneously breathing extremely preterm neonates (24 0/7 to 27 6/7 weeks' gestation) reaching failure criteria for continuous positive airway pressure within the first 24 hours of life. Data were analyzed from April 2023 to January 2024. Intervention: Infants were randomly assigned (1:1) to IN-REC-SUR-E or IN-SUR-E and then followed up. Main outcomes and measures: The primary outcome was the occurrence of death after discharge or major disability at cPNA 2 years. Secondary outcomes were neurodevelopmental outcomes (major disability, cerebral palsy, cognitive impairment, visual deficit, or auditory deficit), anthropometric measurements (weight, length, and head circumference), and recurrent respiratory infections and hospitalizations because of respiratory causes at 2y cPNA. Results: A total of 137 extremely preterm infants (median [IQR] gestational age, 26.5 [25.3-27.5] weeks and 75 [54.7%] female), initially enrolled in the original RCT, were followed up at cPNA 2 years, including 64 infants in the IN-SUR-E group and 73 infants in the IN-REC-SUR-E group. There were no significant differences in the occurrence of death after discharge or major disability at cPNA 2 years (IN-SUR-E: 13 children [20.3%] vs IN-REC-SUR-E: 10 children [13.7%]; P = .36). There were no significant differences in incidence of disability, cerebral palsy, or cognitive impairment in the IN-REC-SUR-E group compared with the IN-SUR-E group. There were no significant differences in anthropometric measurements (weight, length, and head circumference) between groups. There were no significant differences in the incidence of recurrent respiratory infections or in hospitalizations because of respiratory causes between groups. Conclusions and relevance: In this RCT of lung recruitment before surfactant vs standard care there were no significant differences between the 2 groups in death, neurodevelopmental outcomes, anthropometric measurements, or recurrent respiratory infections at the 2-year follow-up. These findings can aid clinicians in decision-making for the best strategy to administer surfactant, considering long-term outcomes.

PMID: 39320892

27. Antenatal magnesium sulphate reduces cerebral palsy after preterm birth, implementation into clinical practice needs to be accelerated globally to benefit preterm babies

Karen Luyt

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No abstract available

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