1. Strength Training for Adolescents with cerebral palsy (STAR): study protocol of a randomised controlled trial to determine the feasibility, acceptability and efficacy of resistance training for adolescents with cerebral palsy.


INTRODUCTION: Gait is inefficient in children with cerebral palsy, particularly as they transition to adolescence. Gait inefficiency may be associated with declines in gross motor function and participation among adolescents with cerebral palsy. Resistance training may improve gait efficiency through a number of biomechanical and neural mechanisms. The aim of the Strength Training for Adolescents with cerebral palsy (STAR) trial is to evaluate the effect of resistance training on gait efficiency, activity and participation in adolescents with cerebral palsy. We also aim to determine the biomechanical and neural adaptations that occur following resistance training and evaluate the feasibility and acceptability of such an intervention for adolescents with cerebral palsy. METHODS AND ANALYSIS: 60 adolescents (Gross Motor Function Classification System level I-III) will be randomised to a 10-week resistance training group or a usual care control group according to a computer-generated random schedule. The primary outcome is gait efficiency. Secondary outcomes are habitual physical activity, participation, muscle-tendon mechanics and gross motor function. General linear models will be used to evaluate differences in continuous data between the resistance training and usual care groups at 10 and 22 weeks, respectively. A process evaluation will be conducted alongside the intervention. Fidelity of the resistance training programme to trial protocol will be quantified by observations of exercise sessions. Semistructured interviews will be conducted with participants and physiotherapists following the resistance training programme to determine feasibility and acceptability of the programme.

PMID: 27707836

2. Trunk deformity evaluation based on 3D measurements of front body surface landmarks in people with severe physical disabilities.


PURPOSE: To assess reliability and validity of a trunk deformity evaluation method expressed as rotational and lateral lean angles between the upper and the lower trunk and between the lower trunk and the pelvis using 3D positions of six front body surface landmarks. METHODS: Inter- and intra-rater reliabilities of the proposed method in adults with typical development (n = 22) were assessed, and its validity was also assessed through correlations between the Cobb angle and the analyzed trunk deformity parameters in adults with severe physical disabilities (n = 22). RESULTS: The mean differences between two raters
and between the initial and second measures were within 2°. Moderate correlations were found between the Cobb angle and both the upper and the lower trunk lateral lean angle and the upper trunk rotation angle. CONCLUSIONS: The proposed trunk deformity evaluation appears to be a reliable and valid approach for bedridden people with physical disabilities.

PMID: 27715377

3. Clinical evaluation of ultrasound screening in follow-up visits of infants with cerebral palsy at high risk for developmental dysplasia of the hip.


The aim of the study was to assess the clinical value of ultrasound screenings for the developmental dysplasia of the hip (DDH) and explore its etiology in high-risk infants with cerebral palsy in follow-up visits. A group of 98 cases of infants at high-risk of cerebral palsy who received rehabilitation treatment between July, 2009 and July, 2010 were selected. Infants included 58 men and 40 women, aged <6 months and not lost to follow-up visits. Ultrasound (using Graf static inspection) screening of hips was performed and the results were analyzed and there were 40 abnormal cases among the 98 cases of infants at high risk of cerebral palsy, including 18 cases of unstable hip joint, and 22 cases of DDH (12 cases of hip dysplasia, 3 cases of hip subluxation and 7 cases of hip dislocation). Early clinical intervention for infants with hip dysplasia and outpatient follow-up for infants aged 1-2 years was carried out and had ischemic necrosis of femoral head, with the exception of 1 case of femoral detorsion that was poorly restored. In conclusion, the probability of DDH was higher in infants at high-risk of cerebral palsy compared to the normal infants. Hip ultrasound is a safe, simple, and effective screening method for these infants, which is of great clinical significance for an earlier diagnosis and treatment of DDH in infants with cerebral palsy.

PMID: 27698744

4. Total hip replacement in young non-ambulatory cerebral palsy patients.

Morin C, Ursu C, Delecourt C.


INTRODUCTION: The everyday life of a non-ambulatory adolescent or young adult with cerebral palsy can be severely impaired by a painful or stiff hip. The usual surgical solutions such as proximal femoral resection (PFR) are not entirely satisfactory for pain relief, and are mutilating. HYPOTHESIS: A retrospective study assessed the impact of total hip replacement (THR) on such impairment, on the hypothesis that it is more effective than PFR in relieving pain, without aggravating disability. PATIENTS AND METHODS: The surgical technique consisted in implanting a dual-mobility prosthesis with uncemented acetabular component and cemented femur, after upper femoral shaft shortening and short hip-spica cast immobilization. Forty THRs were performed in 33 patients, including 31 with multiple disability. Follow-up assessment focused on change in functional status, pain, and range of motion. RESULTS: Mean follow-up was 5 years. Pain was more or less entirely resolved. Improvement in range of motion was less striking, and there was no significant change in functional status. There were 2 general, 2 septic and 10 mechanical complications, 6 of which required surgical revision. DISCUSSION: In non-ambulatory cerebral palsy, THR provided much better alleviation of pain than found with PFR treatment. It should be reserved for patients able to withstand fairly long surgery and with femur size compatible with implantation of a femoral component, however small.

PMID: 27697405


This article aims to describe the clinical features of electric powered indoor/outdoor wheelchair (EPIOC) users with cerebral palsy (CP) that are problematic to optimal prescription and to explore comorbidities, features of CP, and conditions secondary to disability impacting on equipment provision for children and adults. The method is a cross-sectional study of EPIOC users (n = 102) with a primary diagnosis of CP. This is a retrospective review of electronic and case note records of EPIOC recipients attending a specialist wheelchair service in 2007-2008. Records were reviewed by a rehabilitation consultant. Data were extracted under three themes; demographic, diagnostic/clinical and wheelchair factors. There were 48 males mean age 27.5 (range 8-70, SD 13.9) years and 54 females, mean age 29.5 (range 7-68, SD 14.6) years with CP. Sixteen comorbidities, nine features of CP, and five features of disability influenced wheelchair prescription. Sixty-four users were provided with specialized seating (SS) and 47 with tilt-in-space (TIS) seats. Complex controls were provided to 16 users, 12 tray-mounted. The majority of users had both SS and TIS. Powered wheelchair prescription has important therapeutic roles in clinical management in addition to enhancing mobility, independence and participation. Clinical features such as spasticity and problematic pain appeared less well managed in adults than in children.

PMID: 27715489

6. [Palsy of the upper limb: Obstetrical brachial plexus palsy, arthrogryposis, cerebral palsy].

[Article in French]

"Palsy of the upper limb" in children includes various diseases which leads to hypomobility of the member: cerebral palsy, arthrogryposis and obstetrical brachial plexus palsy. These pathologies which differ on brain damage or not, have the same consequences due to the early achievement: negligence, stiffness and deformities. Regular entire clinical examination of the member, an assessment of needs in daily life, knowledge of the social and family environment, are key points for management. In these pathologies, the rehabilitation is an emergency, which began at birth and intensively. Splints and physiotherapy are part of the treatment. Surgery may have a functional goal, hygienic or aesthetic in different situations. The main goals of surgery are to treat: joints stiffness, bones deformities, muscles contractures and spasticity, paresis, ligamentous laxity.

PMID: 27692236


Atasavun Uysal S, Baltaci G. Games Health J. 2016 Oct 5. [Epub ahead of print]

OBJECTIVE: This study aimed at assessing how the addition of Nintendo Wii™ (NW) system to the traditional therapy influences occupational performance, balance, and daily living activities in children with spastic hemiplegic Cerebral Palsy (CP). MATERIALS AND METHODS: The present study is a single-blind and randomized trial involving 24 children aged 6-14 years, classified as level I or II on the Gross Motor Function Classification System. The children were allocated into two groups: an intervention and a control group, and their families participated in the study. The activity performance analysis of the children was undertaken by using the Canadian Occupational Performance Measure (COPM), functional balance was measured with the Pediatric Balance Scale (PBS), and activities of daily living were assessed with Pediatric Evaluation of Disability Inventory (PEDI). Twenty-four children with CP were randomly divided into two groups: intervention (n = 12) and...
control group (n = 12). All children in both groups continued their traditional physiotherapy program twice a week, 45 minutes per session, whereas the participants in the intervention group, additionally, were trained with NW, two other days of the week for 12 weeks, with each session lasting for 30 minutes. RESULTS: Self-care, mobility, PEDI total, PBS, and performance of COPM scores increased in the NW group after intervention. Self-care, mobility, and total PEDI increased in the control group as well. However, there was no statistically significant difference found between the groups, except for PBS (P < 0.05).

CONCLUSIONS: NW contributed to the implementation of occupational performance, daily living activities, and functional balance. We recommend that NW could be used in the rehabilitation program to engage play-based activities with fun.

PMID: 27705006

8. Active Videogaming in Youth with Physical Disability: Gameplay and Enjoyment.
Malone LA, Rowland JL, Rogers R, Mehta T, Padalabalanarayan S, Thirumalai M, Rimmer JH.
Games Health J. 2016 Oct 3. [Epub ahead of print]
OBJECTIVE: For active videogaming (AVG) to be a meaningful, health-enhancing physical activity option for youth with physical disability, factors related to game performance and enjoyment must be understood. The objective was to explore associations between quality of gameplay, controller usage, heart rate (HR), physical function, and enjoyment during AVG play in youth with physical disability.

METHODS: Participants (5 girls, 11 boys, mean age 13.8 ± 2.7 years) played four AVGs on three platforms (Nintendo® Wii™, Sony PlayStation3 Move, and Microsoft Xbox® Kinect), across three sessions. Participants' primary means of mobility were manual (n = 13) and power (n = 3) wheelchairs; majority were diagnosed with cerebral palsy or spina bifida. Functional level was assessed using 17 International Classification of Functioning, Disability and Health mobility items. Participants played each AVG for 8 minutes with a 5-minute rest. Quality of gameplay and ability to use controller were recorded on a five-point Likert scale. HR was recorded immediately following each game and participants completed the Physical Activity Enjoyment Scale (PACES). PACES scores were compared across games and correlations were examined among the variables.

RESULTS: PACES scores were significantly greater for Wii Punch-Out compared to Xbox Fitness, Sports Rivals, and Zumba, and for PS3 Sports Champions compared to Xbox Zumba. Higher HR was associated with higher quality of gameplay and a higher PACES score. As quality of gameplay increased, the PACES score increased.

CONCLUSION: Game performance and exercise intensity were positively correlated with AVG enjoyment in youth with physical disability, specifically mobility impairments. Further research is warranted to examine the capacity of AVG play to be an enjoyable health-enhancing activity for individuals with physical disability.

PMID: 27696899

9. This month’s summaries focus on approaches to improving quality of life for children with cerebral palsy.
[No authors listed]
Aim To systematically review the impact of different interventions on quality of life (QoL) for children with cerebral palsy.

PMID: 27710252

Mäenpää H, Autti-Rämö I, Varho T, Forsten W, Haataja L.
AIM: To develop a national consensus on outcome measures that define functional ability in children with cerebral palsy (CP) according to the International Classification of Functioning, Disability and Health (ICF) framework. METHOD: The project started in 2008 in neuropaediatric units of two university hospitals and one outpatient clinic. Each professional group selected representatives to be knowledge brokers for their own specialty. Based on the evidence, expert opinion, and the ICF
framework, multiprofessional teams selected the most valid measures used in clinical practice (2009-2010). Data from 269 children with CP were analysed, classified by the Gross Motor Function Classification System, Manual Ability Classification System, and Communication Function Classification System, and evaluated. RESULTS: The process aimed at improving and unifying clinical practice in Finland through a national consensus on the core set of measures. The selected measures were presented by professional groups, and consensus was reached on the recommended core set of measures to be used in all hospitals treating children with CP in Finland. INTERPRETATION: A national consensus on relevant and feasible measures is essential for identifying differences in the effectiveness of local practices, and for conducting multisite intervention studies. This project showed that multiprofessional rehabilitation practices can be improved through respect for and inclusion of everyone involved.

PMID: 27714777


Trabacca A, Vespino T, Di Liddo A, Russo L.


Cerebral palsy (CP) is one of the most frequent causes of child disability in developed countries. Children with CP need lifelong assistance and care. The current prevalence of CP in industrialized countries ranges from 1.5 to 2.5 per 1,000 live births, with one new case every 500 live births. Children with CP have an almost normal life expectancy and mortality is very low. Despite the low mortality rate, 5%-10% of them die during childhood, especially when the severe motor disability is comorbid with epilepsy and severe intellectual disability. Given this life expectancy, children with CP present with a lifelong disability of varying severity and complexity, which requires individualized pathways of care. There are no specific treatments that can remediate the brain damage responsible for the complex clinical-functional dysfunctions typical of CP. There are, however, a number of interventions (eg, neurorehabilitation, functional orthopedic surgery, medication, etc) aimed at limiting the damage secondary to the brain insult and improving these patients' activity level and participation and, therefore, their quality of life. The extreme variability of clinical aspects and the complexity of affected functions determine a multifaceted skill development in children with CP. There is a need to provide them with long-term care, taking into account medical and social aspects as well as rehabilitation, education, and assistance. This long-term care must be suited according to children's developmental stage and their physical, psychological, and social development within their life contexts. This impacts heavily on the national health systems which must set up a network of services for children with CP, and it also impacts heavily on the family as a whole, due to the resulting distress, adjustment efforts, and changes in quality of life. This contribution is a narrative review of the current literature on long-term care for children with CP, aiming at suggesting reflections to improve these children's care.

PMID: 27703369

12. The International Classification of Functioning (ICF) to evaluate deep brain stimulation neuromodulation in childhood dystonia-hyperkinesia informs future clinical & research priorities in a multidisciplinary model of care.

Gimeno H, Lin JP.


The multidisciplinary team (MDT) approach illustrates how motor classification systems, assessments and outcome measures currently available have been applied to a national cohort of children and young people with dystonia and other hyperkinetic movement disorders (HMD) particularly with a focus on dyskinetic cerebral palsy (CP). The paper is divided in 3 sections. Firstly, we describe the service model adopted by the Complex Motor Disorders Service (CMDS) at Evelina London Children's Hospital and King's College Hospital (ELCH-KCH) for deep brain stimulation. We describe lessons learnt from available dystonia studies and discuss/propose ways to measure DBS and other dystonia-related intervention outcomes. We aim to report on current available functional outcome measures as well as some impairment-based assessments that can encourage and generate discussion among movement disorders specialists of different backgrounds regarding choice of the most important areas to be measured after DBS and other interventions for dystonia management. Finally, some recommendations for multi-centre collaboration in regards to functional clinical outcomes and research methodologies for dystonia-related interventions are proposed.

PMID: 27707656

Monbaliu E, De Cock P, Mailleux L, Dan B, Feys H.


AIM: To relate dystonia and choreoathetosis with activity, participation and quality of life (QOL) in children and youth with dyskinetic Cerebral Palsy (CP). METHODS: Fifty-four participants with dyskinetic CP (mean age 14y6m, SD 4y2m, range 6-22y) were included. The Dyskinesia Impairment Scale (DIS) was used to evaluate dystonia and choreoathetosis. Activity, participation and quality of life (QOL) were assessed with the Gross Motor Function Measure (GMFM), the Functional Mobility Scale (FMS), the Jebsen-Taylor Hand Function Test (JTT), the ABILHAND-Kids Questionnaire (ABIL-K), the Life Habits Kids (LIFE-H) and the Quality of Life Questionnaire for children with CP (CP-QOL). Spearman's rank correlation coefficient (rs) was used to assess the relationship between the movement disorders and activity, participation and QOL measures. RESULTS: Significant negative correlations were found between dystonia and the activity scales with Spearman's rank correlation coefficient (rs) varying between -0.65 (95% CI = -0.78 to -0.46) and -0.71 (95% CI = -0.82 to -0.55). Correlations were also found with the LIFE-H (rs = -0.43; 95%CI = -0.64 to -0.17) and the CP-QOL (rs = -0.32; 95%CI = -0.56 to -0.03). As far as choreoathetosis is concerned, no or only weak relationships were found with the activity, participation and quality of life scales. INTERPRETATION: This cross-sectional study is the first to examine the relationship of dystonia and choreoathetosis in dyskinetic CP with the level of activity, participation and QOL. The results revealed dystonia has a higher impact on activity, participation and quality of life than choreoathetosis. These findings seem to suggest it is necessary to first focus on dystonia reducing intervention strategies and secondly on choreoathetosis.

PMID: 27707657


Kuschmann A, Miller N, Lowit A, Pennington L.


PURPOSE: This paper examined the production of intonation patterns in children with developmental dysarthria associated with cerebral palsy (CP) prior to and after speech intervention focussing on respiration and phonation. The study further sought to establish whether intonation performance might be related to changes in speech intelligibility. METHOD: Intonation patterns were examined using connected speech samples of 15 older children with moderate to severe developmental dysarthria due to CP (9 females; age range: 11-18). Recordings were made prior to and after speech intervention based on a systems approach. Analyses are focussed on the use of intonation patterns, pitch accentuation and phrasing. RESULT: Group analyses showed a significant increase in the use of rising intonation patterns after intervention. There were also some indications that this increase might have been related to gains in speech intelligibility for some of the children. No changes were observed regarding pitch accentuation and phrasing following intervention. CONCLUSION: The findings highlight that changes can occur in the use of intonation patterns in children with dysarthria and CP following speech systems intervention. It is hypothesised that the emergence of the rising pattern in some of the children's intonational inventories possibly reflected improved breath support and control of laryngeal muscles.

PMID: 27705032

15. Therapeutic effects of intensive voice treatment (LSVT LOUD) for children with spastic cerebral palsy and dysarthria: A phase I treatment validation study.

Boliek CA, Fox CM.


PURPOSE: The aim of the present study was to validate and extend the evaluation of treatment outcomes following LSVT LOUD® in children with dysarthria secondary to cerebral palsy (CP). METHOD: Seven children (5 females, 6-10 years) with spastic quadriplegia and dysarthria received LSVT LOUD. Outcomes included: (a) quantitative and qualitative indices of
communication and social functioning representing therapeutic effects and (b) features of the acoustic signal representing physiological effects on the speech mechanism. A matched group of typically developing children served as controls. Testing occurred just prior to (PRE), immediately following (POST), and at 12 weeks post-treatment (FUP). RESULT: Expert listeners preferred voice quality and articulatory precision of children with CP at FUP as compared to PRE. Acoustic data indicated improvements on select measures of vocal functioning at POST with some maintenance at FUP. Single word intelligibility improved immediately POST, but was not maintained at FUP. Parents rated positive changes in characteristics of voice and speech and qualitative changes in communication at both POST and FUP. CONCLUSION: The present study validated some of the previous LSVT LOUD outcomes in children with dysarthria and CP and extended our understanding of therapeutic effects through qualitative data obtained from extensive parent interviews.

PMID: 27705010


Voulgarakis H, Forte S.


Pediatric feeding disorders are common among children with developmental disabilities and can have detrimental effects on growth and development. An escape extinction and negative reinforcement-based approach to treating food refusal was examined in a child with cerebral palsy. A changing criterion treatment design was implemented, which allowed the child to exit the treatment area contingent upon the acceptance and ingestion of a pre-determined number of bites. Food acceptance ranged from one to three bites at baseline and exceeded the pre-set criteria for mastery, at 14 bites during the final intervention phase. The study will contribute to the current literature on negative reinforcement procedures used in the treatment of pediatric feeding problems. The study will contribute to increasing the availability of literature pertaining to pediatric feeding problems among children with complex disabilities such as cerebral palsy. The intervention is brief with components to the treatment package which increases utility and ease of implementation. The study demonstrates the applicability of changing criterion design within clinical settings.

PMID: 27703921

17. Orthodontic treatment and follow-up of a patient with cerebral palsy and spastic quadriplegia.

Çifter M, Cura N.


INTRODUCTION: This report describes the clinical orthodontic management of a patient with spastic quadriplegia and cerebral palsy. Guidelines to overcome difficulties encountered during the treatment period are suggested. METHODS: A 13-year-old boy with cerebral palsy and spastic quadriplegia complained of an undesirable oral appearance because of his malocclusion. He had a Class II molar relationship, with severe maxillary and moderate mandibular anterior crowding. Enamel hypoplasia was apparent on all teeth. He had losses of body function and upper extremity function of 70% and 39%, respectively. His physical limitations necessitated a treatment approach that did not rely on patient-dependent appliances. The treatment plan called for maxillary first premolar extractions, mandibular incisor protrusion, and air rotor stripping. RESULTS: The patient's oral function and esthetic appearance were significantly improved. Aligned dental arches with good occlusion were obtained. The patient's self-confidence improved during the treatment period. CONCLUSIONS: Physical appearance can influence personality and social acceptability. Corrective orthodontic treatment for patients with physical handicaps can improve not only oral function, but also self-confidence and self-esteem.

PMID: 27692425
18. Eligibility criteria for therapeutic hypothermia: From trials to clinical practice.

Mehta S, Joshi A, Bajuk B, Badawi N, McIntyre S, Lui K.


AIM: Whole body therapeutic hypothermia (TH) for hypoxic ischaemic encephalopathy was introduced into clinical practice in New South Wales (NSW) and Australian Capital Territory in 2007. State-wide policy adopting the eligibility criteria and practice based on trial-designs was published in 2009. METHODS: The study was conducted by retrospectively reviewing medical records of all TH infants born between 2007 and 2011 in NSW and Australian Capital Territory to examine if eligibility criteria (assessed against evidence-based policy directives) were met. RESULTS: A total of 207 infants received TH, 104 (50%) did not meet the eligibility criteria defined in NSW policy directive. Over the 5-year period, the proportion of infants meeting the eligibility criteria did not change. Seventy percent of infants (73 out of 104) not meeting eligibility criteria did not fulfil the criteria for 'evidence of asphyxia', although half of them met 'moderate or severe encephalopathy criterion'. Adverse events (hypotension, coagulopathy and arrhythmia), were more common in the 'criteria met' group than the 'criteria not met' group (89 vs. 71%, P = 0.001). Similar proportions of infants had TH discontinued before 72 h (criteria met: 32 (31%) vs. criteria not met: 27(26%)). Most frequent reason for early cessation was 'palliation' (19/32, 59%) in criteria met and 'clinical improvement' (16/27, 59%) in criteria not met group. CONCLUSIONS: Many TH infants were treated based on clinician judgement, though not meeting the trial-design policy criteria. Early TH cessation (<72 h) was common. Future studies are warranted on long-term neurodevelopmental outcomes for all infants receiving TH particularly those with early cessation of therapy.

PMID: 27701803

19. The Baby Moves prospective cohort study protocol: using a smartphone application with the General Movements Assessment to predict neurodevelopmental outcomes at age 2 years for extremely preterm or extremely low birthweight infants.


INTRODUCTION: Infants born extremely preterm (EP; <28 weeks' gestation) and/or with extremely low birth weight (ELBW; <1000 g birth weight) are at increased risk for adverse neurodevelopmental outcomes. However, it is challenging to predict those EP/ELBW infants destined to have long-term neurodevelopmental impairments in order to target early intervention to those in most need. The General Movements Assessment (GMA) in early infancy has high predictive validity for neurodevelopmental outcomes in preterm infants. However, access to a GMA may be limited by geographical constraints and a lack of GMA-trained health professionals. Baby Moves is a smartphone application (app) developed for caregivers to video and upload their infant's general movements to be scored remotely by a certified GMA assessor. The aim of this study is to determine the predictive ability of using the GMA via the Baby Moves app for neurodevelopmental impairment in infants born EP/ELBW. METHODS AND ANALYSIS: This prospective cohort study will recruit infants born EP/ELBW across the state of Victoria, Australia in 2016 and 2017. A control group of normal birth weight (>2500 g birth weight), term-born (≥37 weeks' gestation) infants will also be recruited as a local reference group. Parents will video their infant's general movements at two time points between 3 and 4 months' corrected age using the Baby Moves app. Videos will be scored by certified GMA assessors and classified as normal or abnormal. Parental satisfaction using the Baby Moves app will be assessed via survey. Neurodevelopmental outcome at 2 years' corrected age includes developmental delay according to the Bayley Scales of Infant and Toddler Development-III and cerebral palsy diagnosis.

PMID: 27697883

van Karnebeek CD, Bowden K, Berry-Kravis E.


BACKGROUND: Neurogenetic developmental conditions represent a heterogeneous group of rare inherited disorders with neurological manifestation during development. Treatments for these conditions have largely been supportive; however, a number of treatments are emerging which target the underlying physiology and offer great potential. Our aim was to present a state-of-the-art overview of the current and potential causal treatments available or under development for neurogenetic developmental conditions. METHODS: In this review, we focus on the following neurogenetic developmental conditions: (1) inborn errors of metabolism causing neurogenetic developmental conditions, (2) fragile X syndrome, (3) Rett syndrome, (4) tuberous sclerosis complex, 5) Down syndrome and other neurogenetic developmental conditions. RESULTS: A large group of inborn errors of metabolism leads to neurodevelopmental disability, affecting the central nervous system during infancy or childhood and can present with comorbidities such as intellectual developmental disability, epilepsy, atypical cerebral palsy, autism spectrum disorder, behavioral and psychiatric disturbances, for which causal treatments are discussed. CONCLUSIONS: The advent of these new disease-modifying therapies has the potential to reverse the underlying neural mechanisms of these debilitating conditions, which may provide prospect to affected individuals.

PMID: 27697313