Litteraturgjennomgang om barn og unge med Down syndrom

Retningslinje for oppfølging av barn og unge med Down syndrom ble publisert i februar 2017. Det er nå gjort en gjennomgang av ny litteratur med tanke på om det er behov for revidering av anbefalingene i retningslinjen på bakgrunn av ny kunnskap.

Konklusjon etter litteraturgjennomgang
Gjennomgang av ny litteratur avdekker ikke behov for å endre anbefalingene i retningslinjen fra 2017. Artiklene som ble identifisert gjennom de nye litteratursøkene bidrar med kunnskap som styrker kunnskapsgrunnlaget retningslinjen bygger på, og avdekker at det fortsatt er behov for mer forskning innenfor flere av temaene retningslinjen omhandler. En ny litteraturgjennomgang vil bli gjennomført om ca. 3 år.

Metodebeskrivelse
Litteratursøk ble gjennomført i september og oktober 2020 og deretter et oppdatert søk i mars 2022. Søkene ble gjort av bibliotekar ved Medisinsk bibliotek, Oslo Universitetssykehus, Ullevål og Universitetet i Oslo.

Det ble søkt etter litteratur i følgende databaser: Ovid MEDLINE, APA PsycInfo, PubMed, Cochrane og Epistemonikos. Det ble tatt utgangspunkt i søkeordene brukt i litteratursøkene som ligger til grunn for retningslinjen. Detaljert informasjon om søkeord og dokumentasjon av søkene kan fås ved henvendelse til RHABU.

I søket foretatt høsten 2020 ble artikler publisert fra og med 2014 og frem til september 2020 inkludert. I det oppdaterte søket som ble gjort i mars 2022 ble artikler publisert fra og med oktober 2020 og frem til mars 2022 inkludert.

Det første søket resulterte i 1743 artikler. Kun oversiktsartikler ble inkludert. Etter fjerning av duplikater gjensto 112 oversiktsartikler for nærmere gjennomgang. Disse ble vurdert med tanke på relevans for temaene som retningslinjen omhandler. Etter denne gjennomgangen gjensto 31 oversiktsartikler. Det andre søket resulterte i 495 artikler, hvor av 34 var oversiktsartikler. Etter nærmere vurdering med tanke på relevans for temaene som retningslinjen omhandler gjensto 9 oversiktsartikler.

To personer har hver for seg vurdert artiklene etter gitte kriterier. Der vurderingen ikke var sammenfallende drøftet man seg frem til enighet.

Det ble også gjort et søk etter retningslinjer og kliniske oppslagsverk. Det ble ikke identifisert noen nye retningslinjer for oppfølging av barn og unge med Down syndrom.

I oversikten under presenteres artiklene tematisk. Temaene er satt opp i samme rekkefølge som i Retningslinje for oppfølging av barn og unge med Down syndrom. Artiklene presenteres med årstall for publisering, tittel, forfatter og konklusjon. I tillegg er det satt inn lenke til alle artiklene.

SAMSPILL, KOMMUNIKASJON OG SPRÅK

2022

Title: A systematic review of speech, language and communication interventions for children with Down syndrome from 0 to 6 years

Authors: Seager, E.; Sampson, S.; Sin, J.; Pagnamenta, E.; Stojanovik, V.;

Conclusions: The findings from this review suggest that interventions that have high dosage, focus on language and communication training within a naturalistic setting, and are co-delivered by parents and clinicians/researchers may have the potential to provide positive outcomes for children with DS between 0 and 6 years of age. Due to the limited number of studies, limited heterogeneous data and the moderate to high risk of bias across studies, there is an urgent need for higher quality intervention studies in the field to build the evidence base.

A systematic review of speech, language and communication interventions for children with Down syndrome from 0 to 6 years - PubMed (nih.gov)

2020

Title: Children with Down syndrome can benefit from language interventions; Results from a systematic review and meta-analysis

Authors: Smith, E.; Hokstad, S.; Naess, K. B.;

Conclusion: The findings from the present study suggest that providing interventions aiming to improve language for children with Down syndrome is more effective than business as usual or another comparison intervention. The Hawthorne effect (see Gast, 2010) could contribute to this outcome to some extent, where additional attention and new energy invested toward the child's language activities may motivate and encourage both the implementer and the child. The only common component across all the effective studies is the aim to improve language skills, highlighting the need for prioritisation of language interventions in the children's curriculum and in practice. Additionally, little is known about individual variation in response to interventions. At this time, interventions should therefore be provided to all children with Down syndrome, as responsiveness cannot be predicted

2018

Title: Augmentative and alternative communication in children with Down's syndrome: a systematic review

Authors: Barbosa, R. T. A.; de Oliveira, A. S. B.; de Lima Antao, J. Y. F.; Crocetta, T. B.; Guarnieri, R.; Antunes, T. P. C.; Arab, C.; Massetti, T.; Bezerra, I. M. P.; de Mello Monteiro, C. B.; de Abreu, L. C.;

Conclusion: Twelve instruments that significantly aided in the communication and socialization of children with DS were identified from this review. This study highlights that these instruments provide significant results for children with DS not only in terms of their interaction with each other but also their interactions with other people who coexist with this population, thereby improving interpersonal relationships. However, some key factors should be considered in using such

technological devices, including preferences, professional and parent training, joint use of the devices, display design, and above all stratification of the cognitive level before any intervention. Future investigations in communication and socialization of children with DS should employ standardized methods.

Augmentative and alternative communication in children with Down's syndrome: a systematic review - PubMed (nih.gov)

2018

Title: Communication intervention for individuals with Down syndrome: Systematic review and meta-analysis

Authors: Neil, N.; Jones, E. A.;

Conclusion: The results suggest that behavior analytic strategies are a promising approach, and future research should focus on replicating the effects of these interventions with greater methodological rigor.

Communication intervention for individuals with Down syndrome: Systematic review and meta-analysis - PubMed (nih.gov)

KOGNISJON

2020

Title: The relationship between chronic health conditions and cognitive deficits in children, adolescents, and young adults with down syndrome: A systematic review

Authors: Gandy, K. C.; Castillo, H. A.; Ouellette, L.; Castillo, J.; Lupo, P. J.; Jacola, L. M.; Rabin, K. R.; Raghubar, K. P.; Gramatges, M. M.;

Conclusion: Individuals with Down syndrome exhibit deficits in cognitive ability, particularly related to attention, executive function and verbal processing. These deficits may be further exacerbated by the presence of chronic health conditions, particularly sleep disorders. Individuals with Down syndrome and co-occurring sleep disorders may benefit from early interventions to mitigate their risk for adverse cognitive outcomes.

The relationship between chronic health conditions and cognitive deficits in children, adolescents, and young adults with down syndrome: A systematic review - PMC (nih.gov)

2020

Title: Systematic Review of Recent Pediatric Down Syndrome Neuropsychology Literature: Considerations for Regression Assessment and Monitoring

Authors: Grealish, Katherine G.; Price, August M.; Stein, D.S.

Conclusion: Findings argued against a single “DS profile” and revealed multiple within-group differences as well as expected and unexpected differences relative to typically developing children and children with other intellectual and developmental disabilities. Areas identified as most germane

to regression monitoring included working memory, inhibition, letter and word identification, navigational route learning, motor skills (when strong at baseline), single word receptive/expressive vocabulary, and adaptive function

Systematic Review of Recent Pediatric Down Syndrome Neuropsychology Literature: Considerations for Regression Assessment and Monitoring - PubMed (nih.gov)

2018

Title: Memory profiles in Down syndrome across development: a review of memory abilities through the lifespan

Authors: Godfrey, M.; Lee, N. R.;

Conclusion: In conclusion, while behavioral research studies and neuroimaging investigations have provided a great deal of insight into the nature of memory impairments in DS and their neural correlates, more functional imaging and longitudinal research is needed to examine behavior-brain relations directly in DS. In addition, research employing nuanced memory tasks that have analogues for murine models of DS will be important for advancing our understanding of the neural and possibly genetic underpinnings of the complex memory difficulties faced by individuals with DS. Such research could shed light on the etiology and developmental unfolding of these impairments and their (predictive) relations to real-world functioning over time. In turn, these investigations may support the development of targeted educational and pharmacological interventions to improve quality of life for those with DS and their families.

Finally, it is important to note that the quality of life and lifespan trajectories of individuals with DS have significantly improved over the past several decades and will hopefully continue to do so. Consequently, our understanding of adults with DS today cannot be used to predict the well-being and capabilities of an adult with DS in future years. Thus, it is important to recognize that descriptions of adults with DS are evolving, and thus, research will continue to be needed to provide more accurate descriptions of the capabilities and needs of adults with DS in future generations.

Memory profiles in Down syndrome across development: a review of memory abilities through the lifespan | Journal of Neurodevelopmental Disorders | Full Text (biomedcentral.com)

2018

Title: Joint attention in Down syndrome: A meta-analysis

Authors: Hahn, L. J.; Loveall, S. J.; Savoy, M. T.; Neumann, A. M.; Ikuta, T.;

Conclusion: The present study represents the first meta-analysis, to our knowledge, of joint attention in DS. Even though there are very few studies on this topic, the results provide more conclusive evidence than was previously available that joint attention is a relative strength in DS, especially when compared to individuals with other DDs. The presence of strengths in joint attention have implications for early intervention efforts for this population, especially for language development. More research is needed to examine correlates of within-syndrome variability and the longitudinal influence, or cascading effects, of joint attention on other domains of development.

Joint attention in Down syndrome: A meta-analysis - ScienceDirect

MOTORIKK

2021

Title: Gait characteristics and effects of early treadmill intervention in infants and toddlers with down syndrome: a systematic review

Authors: Kinaci-Biber, E.; Onerge, K.; Mutlu, A.;

Conclusion: Toddlers with DS and TD show similar improvement in spatiotemporal parameters, but the reduction in step width is not sufficient in DS. Early treadmill training has a positive effect on DS infants by providing an increase in stride length and a decrease in stride width in gait characteristics which clinicians should consider in treatment and rehabilitation protocols. However, studies that show the efficacy of treadmills are needed to compare TD peers with toddlers with DS who receive early treadmill training. It should also be investigated why kinetic and sEMG data are not used, especially in intervention studies, in order to clarify the gait patterns seen in infants with DS and to plan appropriate intervention.

Gait characteristics and effects of early treadmill intervention in infants and toddlers with down syndrome: a systematic review - PubMed (nih.gov)

2021

Title: Efficacy of Lower Extremity Cycling Interventions for Youth with Intellectual Disabilities: A Systematic Review

Authors: Maria A. Fragala-Pinkham, Amy L. Ball & Lynn M. Jeffries

Conclusion: The evidence on cycling interventions for youth with ID is limited. Short-term cycling interventions may be effective in teaching youth with ID how to ride a two-wheeled bicycle. Therapists can use this information in the clinic setting but should carefully measure outcomes when providing short-term interventions focused on teaching children with ID how to ride a two-wheeled bicycle independently. Children who do learn to ride independently may demonstrate increased physical activity levels and improved body composition one-year after learning how to ride. Weak evidence suggests that cycling may have short-term effects on cognition. Additional research is needed for therapists to make clinical decisions and recommendations related to dosing of cycling interventions. Future studies should measure long-term outcomes which focus on participation, specifically participants' ability to ride a bike in the community with family or friends or for transportation to school, work or recreational activities.

Efficacy of Lower Extremity Cycling Interventions for Youth with Intellectual Disabilities: A Systematic Review - PubMed (nih.gov)

2020

Title: Postural control in Down syndrome and relationships with the dimensions of the International Classification of Functioning, Disability and Health – a systematic review

Authors: Brugnaro, B. H.; Oliveira, M. F. P.; de Campos, A. C.; Pavao, S. L.; Rocha, Nacf;

Conclusion: There are a large number of studies addressing postural control in children and adolescents with Down syndrome, which report that they show decreased postural stability and greater vulnerability to sensory changes than their typical peers. Nevertheless, we did not find any studies with research designs based on the biopsychosocial model of health proposed by ICF. Although the majority of studies addressed components from the Body Structures and Function and Activity domains in their design, most of them did not associate the activity level of the participants with postural control. Moreover, none of them included an evaluation of

Participation and Environmental Factors, which are potential factors impacting postural control and functioning. Moreover, only a few studies were classified as good, which limits the translation of results to clinical practice. Thus, further studies addressing postural control in this population should, as much as possible, use the ICF framework to characterize postural control, thus emphasizing the functional relevance of this motor ability.

Postural control in Down syndrome and relationships with the dimensions of the International Classification of Functioning, Disability and Health - a systematic review - PubMed (nih.gov)

2020

Title: Systematic Review of the Main Motor Scales for Clinical Assessment of Individuals with down Syndrome

Authors: Moriyama, C. H.; Massetti, T.; Crocetta, T. B.; Silva, T. D. D.; Mustacchi, Z.; Guarnieri, R.; De Abreu, L. C.; Araujo, A. V. L.; Menezes, L. D. C.; Monteiro, C. B. M.; Leone, C.

Conclusion: Although the identified scales have not been frequently used in different studies, the motor scales found in the present review can be used by researchers and health professionals, particularly considering their demonstrated reliability, sensitivity and specificity. However, further studies are needed to establish which scales are most sensitive for motor assessment of the Down syndrome population. These scales, except the GMFM[19,34], were designed for the evaluation of the development of typical children, but due to their validity and reliability have been applied to children with DS to follow their evolution and compare them with themselves. The professionals must know the instrument that they intend to use so that the use meets the established objectives.

Also, tests that can be used for the elderly population with DS have been indicated, taking into account that the motor difficulties due to aging may impair their participation in their social environment.

Systematic Review of the Main Motor Scales for Clinical Assessment of Individuals with down Syndrome - PubMed (nih.gov)

2018

Title: Interventions to improve sensory and motor outcomes for young children with central hypotonia: A systematic review

Authors: Paleg, G.; Romness, M.; Livingstone, R.;

Conclusion: Green light evidence supports treadmill training (to promote ambulation and gait characteristics) and massage (to positively affect muscle tone, motor development and use of vision)

for infants with Down syndrome. These interventions are considered Yellow (possibly effective) for other populations. Green light evidence supports impact of orthoses on foot alignment for ambulatory children with hypotonia, while impact on gait characteristics is Yellow light and motor development may be negatively impacted (Red light) in pre-ambulatory children. All other interventions rated Yellow (possibly effective) and therapists should monitor using sensitive outcome measures.

Interventions to improve sensory and motor outcomes for young children with central hypotonia: A systematic review - IOS Press

2018

Title: Effects of whole-body vibration on muscle strength, bone mineral content and density, and balance and body composition of children and adolescents with Down syndrome: a systematic review

Authors: Saquetto, M.B,; Pereira, F.F.; da Silva, C.M.; Conceicao, C.S.; Gomes Neto, M.;

Conclusion: The studies included in this systematic review showed that WBV training has positive effects on bone mineral density (BMD), body composition and balance. Results of this study showed that WBV training improves muscle strength, BMD, body composition and balance of children and adolescents with Down syndrome, and a more in-depth analysis of its effects on other variables in this population is required, as well as of parameters to be used

Effects of whole-body vibration on muscle strength, bone mineral content and density, and balance and body composition of children and adolescents with Down syndrome: a systematic review - PubMed (nih.gov)

2017

Title: Musculoskeletal development in patients with Down syndrome: Review article

Authors: Dupre, Corey MPAS, PA-C; Weidman-Evans, Emily PharmD, BC-ADM

Conclusion: Down syndrome causes musculoskeletal deficits that disrupt patients' ability to develop properly and maintain their activities of daily living. These deficits can be overcome by using calcium supplementation along with exercise programs that focus on gait correction, agility, balance, and muscle strength. These regimens should be started early in life to prevent developmental deficiencies and should be maintained to avoid musculoskeletal injuries later in life

Musculoskeletal development in patients with Down syndrome - PubMed (nih.gov)

2017

Title: Treadmill interventions in children under six years of age at risk of neuromotor delay:

Update of review published in 2011

Authors: Valentín-Gudiol M, Mattern-Baxter K, Girabent-Farrés M, Bagur-Calafat C, Hadders-Algra M, Angulo-Barroso RMaria

Conclusion: This update of the review from 2011 provides additional evidence of the efficacy of treadmill intervention for certain groups of children up to six years of age, but power to find significant results still remains limited. The current findings indicate that treadmill intervention may accelerate the development of independent walking in children with Down syndrome and may accelerate motor skill attainment in children with cerebral palsy and general developmental delay. Future research should first confirm these findings with larger and better designed studies, especially for infants with cerebral palsy and developmental delay. Once efficacy is established, research should examine the optimal dosage of treadmill intervention in these populations

Treadmill interventions in children under six years of age at risk of delay in motor skills | Cochrane

2016

Title: Effects of Neuromuscular Training on Children and Young Adults with Down Syndrome: Systematic Review and Meta-Analysis

Authors: Dai Sugimoto, Samantha L. Bowen, William P. Meehan III , Andrea Stracciolini

Conclusion: Although there were limited studies, the results showed that neuromuscular training could be used as an effective intervention in children and young adults with Down syndrome.

Effects of Neuromuscular Training on Children and Young Adults with Down Syndrome: Systematic Review and Meta-Analysis - PubMed (nih.gov)

SPISEUTVIKLING, ERNÆRING OG VEKST

2021

Title: Can I breastfeed my baby with Down syndrome? A scoping review

Authors: Zhen, L.; Moxon, J.; Gorton, S.; Hook, D.;

Conclusion: In conclusion, this review identified a significant literature gap related to breastfeeding babies with DS. There is insufficient evidence to suggest a difference of barriers and enablers between babies with and without DS. This insufficiency builds a strong foundation for future definitive, high-quality studies to identify these barriers and enablers using pertinent approaches under the current research standards. This review summarised the three main categories of these barriers and enablers. Breastfeeding techniques could improve breastfeeding for babies with DS, although the effectiveness of these techniques needs to be further researched. The need for regular, high-quality professional development and health professional breastfeeding support for mothers of babies with DS is apparent. DS does not prevent babies from breastfeeding, instead maternal patience, determination and support from health professionals can and does make it possible to optimise breastfeeding for babies with DS.

Can I breastfeed my baby with Down syndrome? A scoping review - PubMed (nih.gov)

2020

Title: Nutritional challenges in children and adolescents with Down syndrome: Review article

Authors: Marianne Nordstrøm, Kjetil Retterstøl, Sigrun Hope, Svein Olav Kolset

Conclusion: Increased focus on nutritional measures is important for the health and wellbeing of children and adolescents with Down syndrome. Specific clinical features of Down syndrome have nutritional relevance and need to be addressed systematically. There are different nutritional implications in various age groups, with feeding problems predominant in the first years of life. Clinical screening for feeding problems and evaluation of children with feeding difficulties and low weight gain is important. Excessive weight gain is a concern for many children with Down syndrome aged 4–5 years and above. This concern calls for early prevention to avoid later comorbidities. The switch between preventing the risk of undernutrition in the child's first year of life and obesity in later life is a challenge to treatment. Clearly, a need exists for more research on nutritional aspects in the prevention and treatment of obesity in Down syndrome.

Nutritional challenges in children and adolescents with Down syndrome - PubMed (nih.gov)

2018

Title: Outcome of orthodontic palatal plate therapy for orofacial dysfunction in children with Down syndrome: A systematic review

Authors: F Javed , Z Akram , A P Barillas , S V Kellesarian , H B Ahmed, J Khan, K Almas

Conclution: The effect of OPPT is achieved only in addition to physiotherapy, speech therapy and/or orofacial regulation therapy. Further longitudinal trials with standard evaluation methods, age of children for treatment initiation, treatment duration and standard orofacial variable outcomes are recommended.

Outcome of orthodontic palatal plate therapy for orofacial dysfunction in children with Down syndrome: A systematic review - PubMed (nih.gov)

PSYKISK HELSE

2020

Title: Systematic Review and Meta-analysis: Mental Health in Children With Neurogenetic Disorders Associated With Intellectual Disability

Authors: Emma J Glasson , Nicholas Buckley, Wai Chen, Helen Leonard ,Amy Epstein , Rachel Skoss , Peter Jacoby , A Marie Blackmore , Jenny Bourke , Jenny Downs

Conclusion: Differential vulnerability for psychiatric phenotype expression across the disorders was observed. Syndromes with higher levels of social ability or competence appear to offer relative protection against developing psychopathology. This preliminary finding merits further exploration.

Systematic Review and Meta-analysis: Mental Health in Children With Neurogenetic Disorders Associated With Intellectual Disability - PubMed (nih.gov)

MEDISINSKE OG NEVROLOGISKE TEMA

2020

Title: Multiorgan involvement and management in children with Down syndrome: Review article

Authors: Niamh Lagan, Dean Huggard , Fiona Mc Grane, Timothy Ronan Leahy, Orla Franklin, Edna Roche , David Webb, Aengus O' Marcaigh, Des Cox, Afif El-Khuffash, Peter Greally, Joanne Balfe, Eleanor J Molloy

Conclusion: Children with DS are at an increased risk of multiorgan comorbidities. Organ-specific health surveillance may provide holistic care for the children and families with DS throughout childhood.

Multiorgan involvement and management in children with Down syndrome - PubMed (nih.gov)

ATLANTOAKSIAL INSTABILITET

2018

Title: Iatrogenic neurological injury in children with trisomy 21: Review article

Authors: Renata E. Husnudinov, George M. Ibrahim, Evan J. Propst, Nikolaus E. Wolter,

Conclusion: Iatrogenic neurological injury in children with trisomy 21 are rare but severe and likely under reported. Although the role of preoperative screening remains controversial, all children with trisomy 21 undergoing surgery should be considered at risk for neurological injury due to confirmed or undiagnosed AAI or AOI and should be transferred and positioned with appropriate caution. Children with instability should be referred for neurosurgical attention for preoperative stabilization to mitigate perioperative risk. It is imperative to consider the possibility of neurological injury secondary to medical procedures, as it is clear that neck manipulation of any sort places these children at risk.

Iatrogenic neurological injury in children with trisomy 21 - PubMed (nih.gov)

FERTILITET, GYNEKOLOGI OG PREVENSJON

2019

Title: Management of contraceptives and menstrual complaints in patients with Down syndrome: Review of the literature

Authors: Gustavo Wandresen, Fernanda Sgarbi & Renato Nisihara

Conclusion: There is increasing social inclusion of people with DS. The longer life expectancy and social inclusion of this population mean that medical professionals, especially gynecologists, are increasingly asked to advise on and manage menarche, puberty, menstrual cycles, reproductive health, and sex education for girls and women with DS. Few studies deal with these issues, and there is a shortage of educational material for patients and caregivers and of guidelines and protocols for health-care professionals. Therefore, there is a need for improved medical knowledge about

reproductive health in women with DS. Proper management of menstruation and contraception is an important component of this population's health care. A personalized approach is necessary, geared to the level of patient's intellectual ability and respecting the family's values. It should include appropriate sex education and proper orientation for caregivers. Research is needed to create a structured health-care network for women with DS, enabling them as much as possible to exercise their sexual and reproductive rights.

Management of contraceptives and menstrual complaints in patients with Down syndrome - PubMed (nih.gov)

FORDØYELSE, MAGE OG TARM

2020

Title: Feeding problems and gastrointestinal diseases in Down syndrome: Review article

Authors: A. Ravel, C. Mircher, A.-S. Rebillat, C. Cieuta-Walti, A. Megarbane

Abstract: Feeding problems and gastrointestinal disorders are the most common anomalies in people with Down syndrome (DS) and have a significant impact on their daily life. This study lists the various anomalies on the basis of 504 references selected from a PubMed search in October 2018.

Feeding problems and gastrointestinal diseases in Down syndrome | Elsevier Enhanced Reader

HJERTESYKDOM

2021

Title: Prevalence of pulmonary hypertension among children with Down

syndrome: A systematic review and meta-analysis

Authors: Amar Taksande, Divya Pujari, Patel Zeeshan Jameel, Bharati Taksande, Revat Meshram

Conclusion: This review highlights the increasing prevalence of PH in children with DS. It is crucial for pediatricians to be aware of this morbid disease and channel their efforts towards earlier diagnosis and successful management. Community-based studies with a larger sample size of children with DS should be carried out to better characterize the epidemiology and underlying etiology of PH in DS.

Prevalence of pulmonary hypertension among children with Down syndrome: A systematic review and meta-analysis - PMC (nih.gov)

HOFTEDYSPLASI

2018

Title: Hip Instability in Patients With Down Syndrome: Review article

Authors: Maranho, Daniel MD, PhD; Fuchs, Kathryn MD; Kim, Young-jo MD, PhD; Novais, Eduardo N. MD

Abstract: The incidence of hip instability in children with Down syndrome is 1% to 7%. The natural history is often progressive, with the typical onset of hypermobility of the hip evolving to habitual dislocation, persistent subluxation, and fixed dislocation, and eventually leading to the loss of independent mobility. Treatment focuses on stabilizing the hip joint and depends on the patient's age and the severity of the disease. Typically, surgical intervention is recommended for the treatment of patients with habitual dislocation, subluxation, and complete dislocation of the hip. When indicated, surgical management must take into account associated anatomic abnormalities of the femur and acetabulum. Hip instability in Down syndrome may persist despite surgical intervention and remains a difficult condition to manage.

Hip Instability in Patients With Down Syndrome : JAAOS - Journal of the American Academy of Orthopaedic Surgeons (lww.com)

IMMUNOLOGI

2020

Title: Clinical implications of immune-mediated diseases in children with Down syndrome: Review article

Authors: Ruud H J Verstegen, Krystal J J Chang, Maaike A A Kusters

Conclusion: Individuals with Down syndrome suffer from a variety of immune-mediated conditions that significantly impact their quality of life. While virtually all components of their immune system show abnormalities, finding correlations between these alterations and clinical features remains challenging. In clinical care for individuals with Down syndrome, one should understand the complexity of this condition and the many immunological and non-immunological factors that contribute to a higher risk for (severe) infections. Also, because malignancies, autoimmune and inflammatory conditions are more common, one should look out for these in regular follow-up as well. Further research is needed to understand the complex interactions of the immune system in Down syndrome and the correlation with clinical features, as well as studies to further investigate potential treatment and prophylactic therapy options to support the immune system in these individuals.

Clinical implications of immune-mediated diseases in children with Down syndrome - PubMed (nih.gov)

2018

Title: Down Syndrome and the Risk of Severe RSV Infection: A Meta-analysis

Authors: Andrea A. Beckhaus and Jose A. Castro-Rodriguez

Conclusion: This systematic review reveals that children with DS are at a significantly higher risk of having severe RSV infections (higher hospital admission, mortality, LOS, oxygen requirement, ICU admission, need for respiratory support, and additional medication use) than children without DS.

Down Syndrome and the Risk of Severe RSV Infection: A Meta-analysis | American Academy of Pediatrics (aappublications.org)

2018

Title: The evaluation and management of respiratory disease in children with Down syndrome (DS): Review article

Authors: Alsubie, H. S.; Rosen, D.

Conclusion: Children with DS are prone to wide range of respiratory problems that may originate at any level of respiratory tract, as well as in other organ systems. A comprehensive evaluation of is necessary to identify the underlying causes, to prevent short-term morbidity and mortality, and long-term morbidity in these children.

The evaluation and management of respiratory disease in children with Down syndrome (DS) - PubMed (nih.gov)

SØVNFORSTYRRELSER

2022
Title: Management of residual OSA post adenotonsillectomy in children with Down Syndrome: A systematic review

Authors: Ravutha Gounden, M.; Chawla, J. K.;

Conclusion: This review identified several methods for management of residual OSA in children with DS that have been reported in the literature to date. However, these studies have several limitations, which have been highlighted through this review, and there remains a paucity of robust studies in this area. At present it remains unclear as to whether CPAP is the optimal second line therapy for children with DS or whether further surgical options should be considered instead. It is likely that different strategies will be required for different sub-groups within this population and that the development of a phenotypically driven treatment algorithm will be important clinically. Further research is required to support the development of such a model. Reassuringly seven of the eight identified articles were published in the last 4 years, suggesting that this is an evolving area of study.

Management of residual OSA post adenotonsillectomy in children with Down Syndrome: A systematic review - PubMed (nih.gov)

2020
Title: Long Term Continuous Positive Airway Pressure and Non-invasive Ventilation in Obstructive Sleep

Apnea in Children With Obesity and Down Syndrome

Authors: Verhulst, S.;

Conclusion: OSAS in children with obesity and underlying syndromes is highly prevalent. Treatment selection is critical, to limit unsuccessful surgery and in view of the high prevalence of residual OSAS after adenotonsillectomy. Therefore, a high proportion of these children are treated with CPAP or NIV. Overall, PAP therapy has beneficial effects on sleep parameters, daytime symptoms, quality of life, and metabolic parameters. However, these data are generated from a limited number of studies and more studies on the effects of CPAP are certainly warranted. Compliance can be an issue in

obese children and patients with Down syndrome for instance, however an intensive initial follow-up can certainly be helpful in these cases. The use of HFNC certainly deserves more study in CPAP intolerant patients.

Long Term Continuous Positive Airway Pressure and Non-invasive Ventilation in Obstructive Sleep Apnea in Children With Obesity and Down Syndrome - PubMed (nih.gov)

2020
Title: The impact of sleep problems on functional and cognitive outcomes in children with Down syndrome: a review of the literature

Authors: Jasneek K. Chawla, Scott Burgess, Helen Heussler,

Conclusion: The impact of coexisting sleep disorders in children with Down syndrome has not been widely studied, with only 15 relevant studies found through an extensive literature review. Large well-designed studies are required to fully understand this relationship further. This is important as sleep-disordered breathing and difficulties with sleep patterns and routines are highly prevalent in children with Down syndrome. Sleep may be one of the few treatable factors that can assist in improving long-term outcomes in this population.

The impact of sleep problems on functional and cognitive outcomes in children with Down syndrome: a review of the literature - PubMed (nih.gov)

2019
Title: Sleep and sleep disordered breathing in children with down syndrome: Effects on behaviour, neurocognition and the cardiovascular system: Clinical review
Authors: Rosemary SC. Horne, Poornima Wijayaratne, Gillian M. Nixon, Lisa M. Walter

Conclusion: Children with DS have an increased prevalence of sleep problems,including difficulties in initiating and maintaining sleep and excessive daytime sleepiness, compared to TD children. Additionally,the craniofacial abnormalities, obesity and hypotonia that characterise the condition significantly increase their risk of SDBfurther adding to their sleep disturbance. Despite recommendations for early diagnosis and treatment, less than half of the children with DS undergo polysomnography to diagnose the severity of SDB. Evidence to date suggests that the presence of SDB may contribute to behaviour and cognitive problems in these children. Furthermore, children with DS and comorbid SDB have attenuated cardiovascular responses to spontaneous arousals and respiratory events, and a dampened sympathetic response, which may exacerbate the adverse cardiovascular effects of this disorder. The most common treatment for OSA in children with or without DS is AT. Children with DS and comorbid OSA demonstrate a significantly reduced chance of cure following AT compared to TD children, and commonly many children go on to have further treatment such as CPAP. Although the literature is currently limited, children with DS do appear to have less sleepiness, improved quality of life and improved behaviour following AT and/or CPAP. To date, improvements to the cardiovascular sequelae of SDB in children with DS have not been reported. This review has clearly identified that there are a dearth of studies regarding the threshold for treatment of SDB in children with DS and the effectiveness of treatment of SDB in improving neurocognitive, behavioural and cardiovascular outcomes in children with DS. We highlight that these areas require further research to better inform screening and treatment guidelines for children with DS.

Sleep and sleep disordered breathing in children with down syndrome: Effects on behaviour, neurocognition and the cardiovascular system - PubMed (nih.gov)

2018

Title: Prevalence of Obstructive Sleep Apnea in Children With Down Syndrome: A Meta-Analysis

Authors: Chia-Fan Lee, Chia-Hsuan Lee, Wan-Yi Hsueh, Ming-Tzer Lin, Kun-Tai Kang

Conclusion: OSA is highly prevalent in children with Down syndrome. Prevalence of moderate to severe OSA is higher in younger age.

Prevalence of Obstructive Sleep Apnea in Children With Down Syndrome: A Meta-Analysis - PubMed (nih.gov)

2017

Title: Otolaryngologic management of Down syndrome patients: what is new?: Review article

Authors: Ethan C Bassett, Mary F Musso

Summary: Questions remain pertinent to the otolaryngologist regarding the ideal management of children with Down syndrome. Additional studies are necessary, to optimize understanding and treatment of this complex population, in particular as opportunities develop with technological advances.

Otolaryngologic management of Down syndrome patients: what is new? - PubMed (nih.gov)

2017

Title: The Efficacy of Adenotonsillectomy for Obstructive Sleep Apnea in Children with Down Syndrome: A Systematic Review

Authors: Javan Nation, MD and Matthew Brigger, MD, MPH

Conclusion: A 51% reduction in the preoperative apnea-hypopnea index can be expected with the intervention of adenotonsillectomy alone in children with Down syndrome. This information is useful for counseling and managing patient and family expectations. It also serves as a reminder to clinicians to obtain a postoperative sleep study, as many of these patients will need nighttime airway support or secondary sleep surgery.

The Efficacy of Adenotonsillectomy for Obstructive Sleep Apnea in Children with Down Syndrome: A Systematic Review - PubMed (nih.gov)

2017

Title: Adenotonsillectomy outcomes in patients with Down syndrome and obstructive sleep apnea: Review article

Authors: Zachary Farhood, Jonathan W Isley, Adrian A Ong, Shaun A Nguyen, Terence J Camilon, Angela C LaRosa, David R White

Conclusion: There is little objective data in the medical literature addressing T&A efficacy in treating OSA in patients with DS patients. Patients show objective improvement in sleep parameters following T&A for OSA. Adenotonsillectomy should be suggested as a first-line treatment for children with DS and OSA, keeping in mind that monotherapy may be insufficient. Future studies utilizing objective measures are required to further quantify the effect in this patient population.

Adenotonsillectomy outcomes in patients with Down syndrome and obstructive sleep apnea - PubMed (nih.gov)

TYROIDEA FORSTYRRELSER

2018

Title: Thyroid Disorders in Subjects with Down Syndrome: An update: Review article

Author: Nermine H Amr

Conclusion: More understanding of the mechanisms behind thyroid gland dysfunction in DS has evolved over the recent years. There seems to be peculiarities regarding the presentation of autoimmune thyroid disease in DS. The metamorphism of thyroid autoimmunity in DS is common, and warrants careful follow up. The “watchful waiting” strategy is generally becoming more popular for subclinical hypothyroidism, with more frequent testing warranted for this subgroup that represents the majority. There is more evidence regarding the value of radioactive iodine treatment for Graves’ disease. Up till now, there is no uniformly worldwide accepted consensus regarding the frequency of screening after the first year of life, and regarding the TSH cut off value for starting treatment.

Thyroid Disorders in Subjects with Down Syndrome: An Update - PubMed (nih.gov)

URINVEIER

2020

Title: Bladder bowel dysfunction in children with Down’s syndrome: Review article

Authors: Nikita R. Bhatt, Louise Murchison, George Yardy, Milind Kulkarni & Azad B. Mathur

Conclusion: This study reviews the largest cohort of patients with BBD in DS. It is common with serious consequences requiring operative intervention. Usual interventions are unreliable due to poor compliance. Early identification and management protect the renal tract. Regular screening for urogenital anomalies in DS is currently not performed. We recommend a thorough history of bladder function in DS patients to identify these cases early.

Bladder bowel dysfunction in children with Down's syndrome - PubMed (nih.gov)