**Supplementary material**

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| **Supplementary table 1.** Evidence after scoping literature review | | | | | | | | | | | | | | |
|  | **Title** | **Type of publication** | **Authors** | **Year** | ***Journal*** | **#patients** | **DMD** | **BMD** | **Controls** | **Intelligence Cognition** | **Neuropsychol-ogical profile** | **Language** | **Psychiatric comorbidities** | **Behavior** |
| 1 | Evidence for early impairment of verbal intelligence in Duchenne muscular dystrophy | research paper | Marsh G.G. & Munsat T. L | 1974 | *Archives of Disease in Childhood* | 34 | x |  |  | x |  |  |  |  |
| 2 | Intellectual functioning in Duchenne muscular dystrophy: a review | review | Karagan N | 1979 | *Psychological Bulletin* |  | x |  |  | *x* |  |  |  |  |
| 3 | Etiology of intellectual impairment in Duchenne muscular dystrophy | research paper | Al-Qudah, A. A., Kobayashi, J., Chuang, S., Dennis, M., & Ray, P. | 1989 | *Pediatric Neurology* | 4 | x |  |  | *x* |  |  |  |  |
| 4 | Cognitive functions in Duchenne muscular dystrophy: a reappraisal and comparison with spinal muscular atrophy. | research paper | Billard C., Gillet P., Signoret J., et al | 1992 | *Neuromuscular disorders* | 24 | x |  | x | *x* | *x* | *x* |  |  |
| 5 | Reading ability and processing in DMD and Spinal Muscular Atrophy | research paper | Billard D., Gillet P., Barthez M., Hommet C., Bertrand P. | 1998 | *Developmental Medicine & Child Neurology* | 21 | x |  | x | *x* |  | *x* |  |  |
| 6 | Severe cognitive impairment in DMD: obvious clinical indication for Dp71 isoform point mutation screening | research paper | Moizard M.-P., Toutain A., Fournier D., Berret F., Raynaud M., Billard C., Andres C. & Moraine C. | 2000 | *European Journal of Human Genetics* | 12 | x |  |  | *x* |  | *x* |  |  |
| 7 | Loss of Dp140 regulatory sequences is associated with cognitive impairment in dystrophinopathies | research paper | Bardoni A., Felisari G., Sironi M., Comi G., Lai M., Robotti M., Bresolin N | 2000 | *Neuromuscular disorders* | 40 | x | x |  | *x* |  |  |  |  |
| 8 | ﻿Loss of Dp140 dystrophin isoform and intellectual impairment in Duchenne dystrophy. | research paper | Felisari G., Martinelli Boneschi F., Bardoni A., et al | 2000 | *Neurology* | 68 | x |  |  | *x* |  |  |  |  |
| 9 | Poor verbal working memory across intellectual level in boys with DMD | research paper | Hinton V.J., De Vivo D.C., Nereo N.E., Goldstein E., Stern Y. | 2000 | *Neurology* | 92 | x |  | x | *x* | *x* |  |  |  |
| 10 | The neurobiology of duchenne muscular dystrophy: learning lessons from muscle? | narrative review | Blake, DJ., Kroger S. | 2000 | *Trends neuroscience* |  | x |  |  | *x* |  |  |  |  |
| 11 | Complete skipping of exon 66 due to novel mutations of the dystrophin gene was identified in two Japanese families of Duchenne muscular dystrophy with severe mental retardation | research paper | Wibawa T, Takeshima Y, Mitsuyoshi I, Wada H, Surono A, Nakamura H, Matsuo M. | 2000 | *brain development* |  | x |  |  | *x* |  |  |  |  |
| 12 | Selective deficits in verbal working memory associated with a known genetic etiology: the neuropsychological profile of DMD | research paper | Hinton V., De Vivo D.C., Nereo N.E., Goldstein E., Stern Y. | 2001 | *J Int Neuropsychol Soc* | 41 | x |  | x |  | *x* | *x* |  |  |
| 13 | Intelligence and Duchenne muscular dystrophy: full-scale, verbal, and performance intelligence quotients | review | Cotton S., Voudouris N., Greenwood K | 2001 | *Developmental Medicine & Child Neurology* |  | x |  |  | *x* |  |  |  |  |
| 14 | Lesson of the week: late diagnosis of Duchenne's muscular dystrophy presenting as global developmental delay. | case report | Essex C., Roper H | 2001 | *BMJ* |  | x |  |  | *x* |  |  |  |  |
| 15 | Relating familial stress to the psychosocial adjustment of adolescents with Duchenne muscular dystrophy | research paper | Reid DT, Renwick RM. | 2001 | *international journal of rehabilitation research* |  | x |  |  |  |  |  |  | *x* |
| 16 | Psychological characteristics of children suffering from Duchenne muscular dystrophy | research paper | Dubiel J, Rokicki W, Skierska A. | 2002 | *Przegl Lek.* | 39 | x |  | x | *x* |  |  |  |  |
| 17 | Three wishes and psychological functioning in boys with duchenne muscular dystrophy. | research paper | Nereo NE, Hinton VJ. | 2003 | *Journal of development behavioral pedatrics* | 74 | x |  | x |  |  |  |  | *x* |
| 18 | Specific cognitive deficits are common in children with DMD | research paper | Wicksell R.K., Kihlgren M., Melin L. & Eeg-Olofsson O. | 2004 | *Developmental Medicine & Child Neurology* | 20 | x |  | x |  | *x* |  |  |  |
| 19 | Dystrophin deletions and cognitive impairment in Duchenne/Becker muscular dystrophy. |  | Giliberto f., Ferreiro V., Dalamon V. et al | 2004 | *Nerological research* | 126 | x | x |  | *x* |  |  |  |  |
| 20 | Dystrophin deletions and cognitive impairment in Duchenne/Becker muscular dystrophy | research paper | Giliberto F, Ferreiro V, Dalamon V, Szijan I. | 2004 | *Neurological research* | 126 | x | x |  | *x* |  |  |  |  |
| 21 | DMD: alpha-dystroglycan immunoexpression in skeletal muscle and cognitive performance | research paper | Campanario da Silva Pereira C., Kiyomoto B.H., Cardoso R., Oliveira A.S.B. | 2005 | *Arq Neuropsiquiatrica* | 19 | x |  |  | *x* |  |  |  |  |
| 22 | Are males with DMD at risk for reading disabilities? | research paper | Hendriksen J.G.M., Vles J.S.H. | 2006 | *Pediatric Neurology* | 25 | x |  |  | *x* | *x* | *x* |  | *x* |
| 23 | Cognitive impairment in neuromuscular disorders | review | Angelo, M. G. D., Bresolin, N., | 2006 | *Muscle & Nerve* |  | x |  |  |  |  |  |  |  |
| 24 | Language disturbances in a group of participants suffering from Duchenne muscular dystrophy: a pilot study. | research paper | Fabbro F., Marini A., Felisari G | 2007 | *Perceptual and motor skills* | 8 | x |  |  | *x* |  | *x* |  |  |
| 25 | Event-related potentials (P300) and neuropsychological assessment in boys exhibiting DMD | research paper | Della Colleta M.V., Scola R.H., Wiemes G.R.M., Fonseca C.N., Mäder M.J., Freund A.A., Werneck L.C. | 2007 | *Arq Neuropsiquiatrica* | 20 | x |  | x | *x* |  |  |  |  |
| 26 | Evaluation of narrative abilities in patients suffering from DMD | research paper | Marini A., Lorusso M.L., D'Angelo M.G., Civati F., Turconi A.C., Fabbro F., Bresolin N. | 2007 | *Brain and Language* | 21 | x |  | x | *x* | *x* |  |  |  |
| 27 | Verbal and memory skills in males with Duchenne muscular dystrophy | research paper | Hinton, VJ., Fee RJ | 2007 | *Journal devalopmental behavioral pediatrics* | 50 | x |  | x |  | *x* | *x* |  |  |
| 28 | Delayed developmental language milestones in children with Duchenne's muscular dystrophy. | research paper | Cyrulnik S,, Fee R., De Vivo D., et al | 2007 | *The journal of pediatrics* | 130 | x |  | x | *x* | *x* | *x* |  |  |
| 29 | Behavior patterns in Duchenne muscular dystrophy: report on the Parent Project Muscular Dystrophy behavior workshop 8-9 of December 2006, Philadelphia, USA | Meeting report | Poysky J | 2007 | *Neuromuscular disorders* |  | x |  |  |  |  |  |  | *x* |
| 30 | Language disturbances in a group of participants suffering from Duchenne muscular dystrophy: a pilot study. | research paper | Fabbro F, Marini A, Felisari G, Comi GP, D'Angelo MG, Turconi AC, Bresolin N. | 2007 | *Perceptual and motor skills* | 8 | x |  |  | *x* |  | *x* |  |  |
| 31 | Treatment of psychiatric comorbidities in a patient with becker muscular dystrophy. | case report | Chaichana KL, Buffington AL, Brandes M, Edwin D, Lee HB. | 2007 | *psychosomatics* | 1 |  | x |  |  |  |  | *x* |  |
| 32 | Cognitive and adaptive deficits in young children with DMD | research paper | Cyrulnik S.E., Fee R.J., Batchelder A., Kiefel J., Goldstein E., Hinton V.J | 2008 | *Journal of the International Neuropsychological Society* | 20 | x |  |  | *x* | *x* | *x* |  |  |
| 33 | Neuropsychiatric disorders in Males with DMD: frequency Rate of ADHD, ASS and OCD | research paper | Hendriksen J.G.M., Vles J.S.H. | 2008 | *Journal of Child Neurology* | 351 | x |  |  |  |  |  | *x* |  |
| 34 | Duchenne muscular dystrophy: A cerebellar disorder? | review | Cyrulnik S.E., Hinton V.J. | 2008 | *Neuroscience and biobehavioral reviews* |  | x |  |  | *x* | *x* |  |  |  |
| 35 | Developmental and Behavioral Disorders Grown Up. | research paper | Birnkrant J.M., Bennet D.S., Noritz G.H., Birnkrant D.J | 2008 | *Journal of development behavioral pedatrics* |  | x |  |  | *x* |  |  |  | *x* |
| 36 | Cognitive and psychological profile of males with Becker muscular dystrophy | research paper | Young HK, Barton BA, Waisbren S, Portales Dale L, Ryan MM, Webster RI, North KN. | 2008 | *Journal of child neurology* | 24 |  | x |  | *x* | *x* |  |  | *x* |
| 37 | Facilitating family adjustment to a diagnosis of Duchenne muscular dystrophy: April 24-25, 2008, Miami, Florida | research paper | Poysky J, Kinnett K. | 2009 | *Neuromuscular disorders* | 15 | x |  |  |  |  |  |  | *x* |
| 38 | Neurobehavioral characteristics of children with DMD | research paper | Donders J., Taneja C. | 2009 | *Child Neurology* | 22 | x |  | x | *x* | *x* |  |  | *x* |
| 39 | Visuospatial attention distrubance in DMD | research paper | Drummond Soares De Moura M.C., Do Valle L.E.R., Dutra Resende M.B., Conti Reed U. & Osternack Pinto, K. | 2009 | *Developmental Medicine & Child Neurology* | 25 | x |  | x |  | *x* |  |  |  |
| 40 | Activities of daily living (ADL) structure of patients with Duchenne muscular dystrophy, including adults. | research paper | ujiwara T., Tanabe A., Uchikawa K. et al | 2009 | *The Keio Journal of medicine* | 72 | x |  |  |  |  |  |  | *x* |
| 41 | Analysis of Dp71 contribution in the severity of mental retardation trough comparison of Duchenne and Becker patients differing by mutation consequences on Dp71 expression | research paper | Daoud F., Angeard N., Demerre B., Martie I., Benyaou R., Leturcq F., Cossée M., Deburgrave N., Saillour Y., Tuffery S., Urtizberea A., Toutain A., Echenne B., Frischman M., Mayer M., Desguerre I., Estournet B., Réveillère C., Penisson-Besnier., Cuisset JM., Kaplan J.C., Héron D., Rivier F., Chelly J. | 2009 | *Human Molecular Geneticx* | 81 | x | x |  | *x* |  |  |  |  |
| 42 | Psychosocial adjustment in males with Duchenne muscular dystrophy: psychometric properties and clinical utility of a parent-report questionnaire. | research paper | Hendriksen J., Poysky J. Schrans D et al | 2009 | *Journal of pediatric psychology* | 282 | x |  |  |  |  |  |  | *x* |
| 43 | Dystrophin gene mutation and the risk of cognitive impairment in DMD | research paper | Taylor P.J., Betts G.A., Maroulis S., Gilissen C., Pedersen R.L., Mowat D.R., Johnston H.M., Buckley M.F. | 2010 | */* | 62 | x |  |  | *x* |  |  |  |  |
| 44 | Intellectual and Behavioral functioning in a South African Cohort of Boys with DMD | research paper | Donald K.A.M., Mathema H., Thomas K.G.F., Wilmshurst J.M. | 2011 | *Journal of Child Neurology* | 6 | x |  | x | *x* | *x* |  |  |  |
| 45 | Brain metabolite composition in relation to cognitive function and dystrophin mutations in boys with Duchenne muscular dystrophy. | research paper | Kreis R, Wingeier K, Vermathen P., et al | 2011 | *NMR in biomedicine* | 16 | x |  | x | *x* | *x* | *x* |  |  |
| 46 | Neuropsychological impairment and the impact of dystrophin mutations on general cognitive functioning of patients with DMD | research paper | Wingeier K., Giger E., Strozzi S., Kreis R., Joncourt F., Conrad B., Gallati S., Steinlin M. | 2011 | *Journal of clinical neuroscience* | 22 | x |  |  | *x* | *x* | *x* |  |  |
| 47 | Neurocognitive profiles in DMD and gene mutation site | research paper | D'Angelo M.G., Lorusso M.L., Civati F., Comi G.P., Magri F., Del Bo R., Gulgieri M., Molteni M., Turconi A.C., Bresolin N. | 2011 | *Pediatric Neurology* | 42 | x |  | x | *x* | *x* | *x* |  |  |
| 48 | Developmental and behavioral disorders grown up: duchenne muscular dystrophy | narrative review | Birnkrant JM, Bennett DS, Noritz GH, Birnkrant DJ. | 2011 | *Journal of development behavioral pedatrics* |  | x |  |  | *x* |  |  |  | *x* |
| 49 | Attention Deficit hyperactivity disorder and cognitive functioning in DMD: phenotype-genotype correlation | research paper | Pane M., Lombardo M.E., Alfieri P., D'Amico A., Bianco F., Vasco G., Piccini G., Mallardi M., Romeo D.M., Ricotti V., Ferlini A., Gualandi F., Vicari S., Bertini E., Berardinelli A., Mercuri E. | 2012 | *Journal of Pediatrics* | 103 | x |  |  | *x* |  |  | *x* |  |
| 50 | Mental retardation in Duchenne muscular dystrophy | review | Nardes F, Araújo AP, Ribeiro MG | 2012 | *Jornal de pediatria* |  | x |  |  | *x* |  |  |  |  |
| 51 | Neuropsychological profile of duchenne muscular dystrophy | research paper | Perumal A., Rajeswaran J., Nalini A et al | 2013 | *Applied neuropsychology children* | 22 | x |  |  | *x* | *x* |  |  | *x* |
| 52 | Motor and cognitive assessment of Infants and young boys with DMD; results from the muscular dystrophy association DMD clinical research network | research paper | Connolly A.M., et others | 2013 | *Neuromuscular disorders* | 24 | x |  |  | *x* |  |  |  |  |
| 53 | Early neurodevelopmental assessment in Duchenne muscular dystrophy | research paper | Pane M., Scalise R., Berardinelli A., D'Angelo G., Ricotti V., Alfieri P., et al | 2013 | *Neuromuscular disorders* | 81 | x |  |  | *x* |  |  |  |  |
| 54 | Neuropsychological and neurobehavioral functioning in DMD: A review | review | Snow W.M., Anderson J.E., Jakobson L.S. | 2013 | *Neuroscience and biobehavioral reviews* |  | x |  |  | *x* |  |  |  | *x* |
| 55 | Molecular characterization of an X(p21.2;q28) chromosomal inversion in a Duchenne muscular dystrophy patient with mental retardation reveals a novel long non-coding gene on Xq28 | case reports | Tran TH, Zhang Z, Yagi M, Lee T, Awano H, Nishida A, Okinaga T, Takeshima Y, Matsuo M. | 2013 | *Journal of human genetics* |  | x |  |  | *x* |  |  |  |  |
| 56 | Phenotypic heterogeneity and phenotype-genotype correlations in dystrophinopathies: Contribution of genetic and clinical databases | research paper | Humbertclaude V, Hamroun D, Picot MC, Bezzou K, Bérard C, Boespflug-Tanguy O, Bommelaer C, Campana-Salort E, Cances C, Chabrol B, Commare MC, Cuisset JM, de Lattre C, Desnuelle C, Echenne B, Halbert C, Jonquet O, Labarre-Vila A, N'guyen-Morel MA, Pages M, Pepin JL, Petitjean T, Pouget J, Ollagnon-Roman E, Richelme C, Rivier F, Sacconi S, Tiffreau V, Vuillerot C, Béroud C, Tuffery-Giraud S, Claustres M. | 2013 | *Revue Neurologique* |  | x |  |  | *x* |  |  |  |  |
| 57 | One year outcome of Boys with DMD using the Bayley III scales of infant and toddler development | research paper | Connolly A.M., et others | 2014 | *Pediatric Neurology* | 24 | x |  |  | *x* |  |  |  |  |
| 58 | Motor and cognitive delay in Duchenne muscular dystrophy: implication for early diagnosis. | research paper | Mirski K., Crawford T. | 2014 | *The journal of pediatrics* | 179 | x |  |  | *x* |  |  |  |  |
| 59 | Cerebellar-dependent associative learning is preserved in Duchenne muscular dystrophy: a study using delay eyeblink conditioning. | research paper | Schara U., Busse M., Timmann D., et al | 2015 | *PloS one* | 9 | x |  | x |  | *x* |  |  |  |
| 60 | Reading impairment in Duchenne muscular dystrophy: A pilot study to investigate similarities and differences with developmental dyslexia | research paper | Astrea G., Pecini C., Gasperini F., Brisca G., Scutifero M., Bruno C., Santorelli, F.M., Cioni G., Politano L., Chilosi A.M., Battini R. | 2015 | *Research in Developmental Disabilities* | 13 | x |  | x |  | *x* | *x* |  |  |
| 61 | Cognitive and psychological profile of males with Becker muscular Dystrophy | research paper | Young H.K., Barton B.A., Waisbren S., Portales L., Ryan M.M., Webster R.I., North K.N. | 2015 | *Journal of Child Neurology* | 24 |  | x |  | *x* | *x* |  | *x* | *x* |
| 62 | Early neurodevelopmental findings predict school age cognitive abilities in DMD: a longitudinal study | research paper | Chieffo D., Brogna C., Berardinelli A., D'Angelo G., Mallardi M., D'Amico A., Alfieri P., Mercuri E., Pane M. | 2015 |  | 41 | x |  |  | *x* |  |  |  |  |
| 63 | Cognitive and Neurobehavioral Profile in Boys with DMD | research paper | Banihani R., Smile S., Yoon G., Dupuis A., Mosleh M., Snider A., McAdam L. | 2015 | *Journal of Child Neurology* | 59 | x |  |  | *x* | *x* | *x* | *x* |  |
| 64 | Neurodevelopmental , emotional , and behavioural problems in Duchenne muscular dystrophy in relation to underlying dystrophin gene mutations | research paper | Ricotti V., Mandy W., Scoto M., et al | 2015 | *Developmental Medicine & Child Neurology* | 130 | x |  |  | *x* |  |  |  | *x* |
| 65 | BURDEN , PROFESSIONAL SUPPORT , AND SOCIAL NETWORK IN FAMILIES OF CHILDREN AND YOUNG ADULTS WITH MUSCULAR | research paper | Magliano L., Patalano M., Sagliocchi A., et al | 2015 | *Muscle & Nerve* | 502 | x | x |  |  |  |  |  |  |
| 66 | Acetylcholine , GABA and neuronal networks : A working hypothesis for compensations in the dystrophic brain | review | Cohen E,, Quarta E, Fulgenzi G;, et al | 2015 | *Brain Research Bulletin* |  | x | x |  | *x* |  |  |  |  |
| 67 | Prevalence of fatigue, pain, and affective disorders in adults with duchenne muscular dystrophy and their associations with quality of life. |  | Pangalila RF, van den Bos GA, Bartels B, Bergen M, Stam HJ, Roebroeck ME. | 2015 | *Archives of physical medicine and rehabilitation* | 80 | x |  |  |  |  |  |  | *x* |
| 68 | Neuropsychological profile of duchenne muscular dystrophy. | research paper | Perumal AR, Rajeswaran J, Nalini A. | 2015 | *Applied neuropsychology children* | 22 | x |  |  | *x* | *x* |  |  |  |
| 69 | Mismatch Negativity Recording in Children With Duchenne Muscular Dystrophy: A Preliminary Study Integrating Neurophysiological and Neuropsychological Results. | research paper | Filippini M, Guerra A, Negosanti A et al. | 2016 | *Journal of child Neurology* | 14 | x |  | x | *x* | *x* | *x* |  |  |
| 70 | Digit Span Performance in Children with Dystrophinopathy : A Verbal Span or Working Memory Contribution ? | research paper | Leafer E., Fee R., Hinton H. | 2016 | *Journal of the International Neuropsychological Society* | 170 | x | x | x |  | *x* |  |  |  |
| 71 | Learning disabilities in neuromuscular disorders: a springboard for adult life | review | Astrea G., Battini R., Lenzi S., Frosini S., Bonetti S., Moretti E., Perazza S., Santorelli F., Pecini C. | 2016 | *Acta Myologica* |  | x |  |  | *x* |  |  |  |  |
| 72 | Muscle and brain: a dyad with important diagnostic and therapeutic implications. | Letter to the editor | Hendriksen, Vles | 2016 | *Developmental Medicine & Child Neurology* |  | x |  |  | *x* |  |  |  |  |
| 73 | The experiences of patients with Duchenne muscular dystrophy in facing and learning about their clinical conditions. | research paper | Fujino H, Iwata Y, Saito T, Matsumura T, Fujimura H, Imura O. | 2016 | *International journal of qualitative study on health and well-being* | 7 | x |  |  |  |  |  |  | *x* |
| 74 | A Novel Mutation in DMD (c.10797+5G>A) Causes Becker Muscular Dystrophy Associated with Intellectual Disability. | case report | Banihani R, Baskin B, Halliday W, Kobayashi J, Kawamura A, McAdam L, Ray PN, Yoon G. | 2016 | *Journal of development behavioral pedatrics* | 1 | x |  |  | *x* |  |  | *x* |  |
| 75 | DMD and West syndrome. | research paper | Cardas R, Iliescu C, Butoianu N et al | 2017 | *Neuromuscular disorders* | 2 | x |  |  |  |  |  |  |  |
| 76 | Profile of cognitive function in adults with duchenne muscular dystrophy. | research paper | Ueda Y, Suwazono, S., Maedo S et al | 2017 | *Brain and development* | 15 | x |  |  | *x* | *x* |  |  |  |
| 77 | Assessing mental health in boys with Duchenne muscular dystrophy : Emotional , behavioural and neurodevelopmental profile in an Italian clinical sample | research paper | Colombo P., Nobile M., Tesei A., et al | 2017 | *European Journal of Peadiatric Neurology* | 47 | x |  |  | *x* |  |  | *x* | *x* |
| 78 | Predictors of Health-Related Quality of Life in boys with Duchenne muscular dystrophy from six European countries | research paper | Otto C, Steffensen B, Hojberg A, et al | 2017 | *Journal of Neurology* | 321 | x |  |  |  |  |  |  |  |
| 79 | Secondary Conditions Among Males With Duchenne or Becker Muscular Dystrophy. | research paper | Latimer R., Street N., Conway K., et al | 2017 | *Journal of child neurology* |  | x | x |  |  |  |  |  |  |
| 80 | Men with Duchenne muscular dystrophy and end of life planning. | research paper | Abbott D, Prescott H, Forbes K, Fraser J, Majumdar A. | 2017 | *Neuromuscular disorders* | 15 | x |  |  |  |  |  |  | *x* |
| 81 | What explains high life satisfaction in men living with Duchenne muscular dystrophy? A preliminary study to inform psychological intervention. | research paper | Graham CD, Rose MR. | 2017 | *Muscle & Nerve* | 16 | x |  |  |  |  |  |  | *x* |
| 82 | Methylphenidate use in males with Duchenne muscular dystrophy and a comorbid attention-deficit hyperactivity disorder. | research paper | Lionarons D,, Hellebrekers D, Klinkenberg S | 2018 | *European Journal of Peadiatric Neurology* | 10 | x |  |  | *x* | *x* |  |  | *x* |
| 83 | ﻿Implicit learning deficit in children with Duchenne muscular dystrophy: Evidence for a cerebellar cognitive impairment? | research paper | Vicari S. Piccini G, Mercuri E, Et al | 2018 | *Plos One* | 31 | x |  | x |  | *x* |  |  |  |
| 84 | Descriptive Phenotype of Obsessive Compulsive Symptoms in Males With Duchenne Muscular Dystrophy | research paper | Lee A., Buckingham E., Kauer A., et al | 2018 | *Journal of Child Neurology* | 39 | x |  |  |  |  |  | *x* |  |
| 85 | The relationship between deficit in digit span and genotype in nonsense mutation Duchenne muscular dystrophy. | research paper | Thangarajh M., Elfrink G., Trifillis P., et al | 2018 | *Neurology* | 169 | x |  |  |  | *x* |  |  |  |
| 86 | Neurodevelopmental Needs in Young Boys with Duchenne Muscular Dystrophy (DMD): Observations from the Cooperative International Neuromuscular Research Group (CINRG) DMD Natural History Study (DNHS). | research paper | Thangarajh M, Spurney C, Gordisch-Dressman et al | 2018 | *PlOS Currents* | 204 | x |  |  | *x* |  |  | *x* | *x* |
| 87 | Brain-related comorbities in boys and men with Duchenne Muscular Dystrophy: a descriptive study | research paper | Hendriksen, R.G.F., Vles J.S.H., Aalbers M.W., Chin R.F.M., Hendriksen J.G.M. | 2018 | *European Journal of Peadiatric Neurology* | 228 | x |  |  | *x* |  |  | *x* | *x* |
| 88 | Cognitive impairment in neuromuscular diseases: A systematic review | review | Orsini M., Ferreira A.C.A., De assis, A. C. D., Magelhaes T., Teixeira S., Bastos V.H., Marinho V., Oliveira A.B., Freitas M.R.G. | 2018 | *Neurology International* |  | x | x |  | *x* |  |  |  |  |
| 89 | Cognitive profile in Duchenne muscular dystrophy boys without intellectual disability: The role of executive functions. | research paper | Battini R, Chieffo D, Bulgheroni S, Piccini G, Pecini C, Lucibello S, Lenzi S, Moriconi F, Pane M, Astrea G, Baranello G, Alfieri P, Vicari S, Riva D, Cioni G, Mercuri E. | 2018 | *Neuromuscular disorders* | 40 | x |  |  | *x* | *x* |  |  |  |
| 90 | Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan. | guidelines | Birnkrant DJ, Bushby K, Bann CM, Apkon SD, Blackwell A, Colvin MK, Cripe L, Herron AR, Kennedy A, Kinnett K, Naprawa J, Noritz G, Poysky J, Street N, Trout CJ, Weber DR, Ward LM; DMD Care Considerations Working Group | 2018 | *The Lancet Neurology* |  | x |  |  | *x* |  |  | *x* | *x* |
| 91 | Psychosocial Management of the Patient With Duchenne Muscular Dystrophy. | guidelines | Colvin MK, Poysky J, Kinnett K, Damiani M, Gibbons M, Hoskin J, Moreland S, Trout CJ, Weidner N. | 2018 | *Pediatrics* |  | x |  |  | *x* |  |  |  | *x* |
| 92 | Social involvement issues in patients with Becker muscular dystrophy: A questionnaire survey of subjects from a patient registry. | research paper | Mori-Yoshimura M, Mizuno Y, Yoshida S, Minami N, Yonemoto N, Takeuchi F, Nishino I, Murata M, Takeda S, Takahashi Y, Kimura E. | 2018 | *Brain and development* | 125 |  | x |  | *x* |  | *x* | *x* | *x* |
| 93 | Autism spectrum disorders in children affected by Duchenne muscular dystrophy. | research paper | Parisi L, Di Filippo T, Glorioso P, La Grutta S, Epifanio MS, Roccella M | 2018 | *Minerva pediatrica* |  | x |  |  |  |  |  | *x* |  |
| 94 | Psychosocial adjustment and parental stress in Duchenne Muscular Dystrophy | research paper | Gocheva V, Schmidt S, Orsini AL, Hafner P, Schaedelin S, Weber P, Fischer D. | 2019 | *European Journal of Peadiatric Neurology* | 34 | x |  |  |  |  |  |  | *x* |
| 95 | Instruments for the Assessment of Behavioral and Psychosocial Functioning in Duchenne and Becker Muscular Dystrophy; a Systematic Review of the Literature | review | Hellebrekers DMJ, Lionarons JM, Faber CG, Klinkenberg S, Vles JSH, Hendriksen JGM. | 2019 | *Journal of pediatric psychology* |  | x | x |  |  |  |  | *x* | *x* |
| 96 | Psychiatric and neurodevelopmental aspects of Becker muscular dystrophy. | research paper | Mori-Yoshimura M, Mizuno Y, Yoshida S, Ishihara N, Minami N, Morimoto E, Maruo K, Nonaka I, Komaki H, Nishino I, Sekiguchi M, Sato N, Takeda S, Takahashi Y. | 2019 | *Neuromuscular disorders* | 105 |  | x |  | *x* |  |  | *x* | *x* |
| 97 | Relationships between DMD mutations and neurodevelopment in dystrophinopathy. | research paper | Thangarajh M, Hendriksen J, McDermott MP, Martens W, Hart KA, Griggs RC; Muscle Study Group and TREAT-NMD. | 2019 | *Neurology* | 196 | x |  |  |  |  | *x* | *x* | *x* |
| 98 | The NIH Toolbox for cognitive surveillance in Duchenne muscular dystrophy. | research paper | Thangarajh M, Kaat AJ, Bibat G, Mansour J, Summerton K, Gioia A, Berger C, Hardy KK, Wagner KR. | 2019 | *Annals of clinical translational neurology* | 30 | x |  |  | *x* |  |  |  |  |
| 99 | Neurodevelopmental, behavioral, and emotional symptoms common in Duchenne muscular dystrophy. | research paper | Darmahkasih AJ, Rybalsky I, Tian C, Shellenbarger KC, Horn PS, Lambert JT, Wong BL. | 2020 | *Muscle & Nerve* | 700 | x |  |  | *x* | *x* | *x* | *x* | *x* |
| 100 | Combining genetics, neuropsychology and neuroimaging to improve understanding of brain involvement in Duchenne muscular dystrophy - a narrative review. | narrative review | Doorenweerd N. | 2020 | *Neuromuscular disorders* |  | x |  |  | *x* | *x* | *x* | *x* | *x* |
| 101 | Longitudinal follow-up of verbal span and processing speed in Duchenne muscular dystrophy. | research paper | Hellebrekers DMJ, Doorenweerd N, Sweere DJJ, van Kuijk SMJ, Aartsma-Rus AM, Klinkenberg S, Vles JSH, Hendriksen JGM. | 2020 | *European Journal of Peadiatric Neurology* | 28 | x |  |  |  | *x* |  |  |  |
| 102 | Cognitive Deficits in Myopathies | review | Peristeri E, Aloizou AM, Keramida P, Tsouris Z, Siokas V, Mentis AA, Dardiotis E. | 2020 | *International journal of molecular sciences* |  | x |  |  | *x* |  |  |  |  |
| 103 | Duchenne Muscular Dystrophy Successfully Treated with Aripiprazole in a Patient with Autism Spectrum Disorder Symptoms Including Irritability | case report | S, Noda, A, Murakami, S, Kimura, M, Minamiyama, M, Katsuno and S, Kuru | 2021 | *International medicine* | 1 | x |  |  |  |  |  | *x* |  |
| 104 | Longitudinal data of neuropsychological profile in a cohort of Duchenne muscular dystrophy boys without cognitive impairment | research paper | Battini, R., Lenzi, S., Lucibello, S., Chieffo, D., Moriconi, F., Cristofani, P., Bulgheroni, S., Cumbo, F., Pane, M., Baranello, G., Alfieri, P., Astrea, G., Cioni, G., Vicari, S. and Mercuri, E. | 2021 | *Neuromuscular disorders* | 40 | x |  |  |  | *x* |  |  |  |
| 105 | Computational cognitive modeling and validation of Dp140 induced alteration of working memory in Duchenne Muscular Dystrophy | research paper | Tyagi, R., Aggarwal, P., Mohanty, M., Dutt, V. and Anand, A. | 2021 | *Scientific reports* |  | x |  |  |  | *x* |  |  |  |
| 106 | Psychosocial adjustment in adults with Duchenne muscular dystrophy: A pilot study on a shortened parent-report questionnaire. | research paper | Weerkamp PMM, Collin P, Maas RJ, Vermeulen RJ, Klinkenberg S, Hendriksen JGM. | 2022 | *Neuromuscular disorders* | 90 | x |  |  |  |  |  |  | *x* |
| 107 | Determining neurodevelopmental manifestations in Duchenne muscular dystrophy using a battery of brief tests. | research paper | Saito Y, Takeshita E, Komaki H, Nishino I, Sasaki M. | 2022 | *Journal of neurological sciences* | 55 | x |  |  | *x* |  |  | *x* |  |
| 108 | A case of delayed diagnosis of Becker muscular dystrophy due to underlying developmental disorders. | case report | Oda S, Mori-Yoshimura M, Oya Y, Sato N, Nishino I, Takahashi Y. | 2022 | *Brain and development* | 1 |  | x |  | *x* |  |  | *x* |  |
| 109 | Clinical and Molecular Spectrum of Muscular Dystrophies (MDs) with Intellectual Disability (ID): a Comprehensive Overview | review | Mohamadian, M., Rastegar, M., Pasamanesh, N., Ghadiri, A., Ghandil, P. and Naseri, M. | 2022 | *Journal of molecular neurosciences* |  | x |  |  | *x* |  |  |  |  |
| 110 | Dystrophinopathy and the brain: A parent project muscular dystrophy (PPMD) meeting report November 11-12, 2021, New York City, NY | Meeting report | MK, Colvin, N, Truba, S, Sorensen, E, Henricson and K, Kinnett | 2022 | *Neuromuscular disorders* |  | x | x |  | *x* | *x* | *x* | *x* | *x* |
| 111 | The neurocognitive profile of adults with Becker muscular dystrophy in the Netherlands | research paper | Koeks, Z., Hellebrekers, D. M. J., van de Velde, N. M., Alleman, I., Spitali, P., van Duyvenvoorde, H. A., Verschuuren, Jjgm, Hendriksen, J. G. M. and Niks, E. H. | 2022 | *Journal of neuromuscular diseases* | 28 |  | x |  | *x* | *x* |  | *x* | *x* |
| 112 | Assessment of dysarthria with Frenchay dysarthria assessment (FDA-2) in patients with Duchenne muscular dystrophy | research paper | Hijikata, N., Kawakami, M., Wada, A., Ikezawa, M., Kaji, K., Chiba, Y., Ito, M., Fujino, E., Otsuka, T. and Liu, M. | 2022 | *Disability and rehabilitation* | 72 | x |  |  |  |  | *x* |  |  |
| 113 | Cognitive and behavioral functioning in two neurogenetic disorders; how different are these aspects in Duchenne muscular dystrophy and Neurofibromatosis type 1? | research paper | Hellebrekers, D. M. J., van Abeelen, S. A. M., Catsman, C. E., van Kuijk, S. M. J., Laridon, A. M., Klinkenberg, S., Hendriksen, J. G. M. and Vles, J. S. H. | 2022 | *PloS one* | 38 | x |  | x | *x* | *x* |  |  |  |
| 114 | Cognitive profile and neuropsychiatric disorders in Becker muscular dystrophy: A systematic review of literature | review | Ferrero, A. and Rossi, M. | 2022 | *Neuroscience and biobehavioral reviews* |  |  | x |  | *x* |  |  | *x* | *x* |
| 115 | Neuropsychological and behavioral profile in a cohort of Becker muscular dystrophy pediatric patients | research paper | Cumbo, F., Tosi, M., Catteruccia, M., Diodato, D., Nicita, F., Capitello, T. G., Alfieri, P., Vicari, S., Bertini, E. and D'Amico, A. | 2022 | *Neuromuscular disorders* | 22 |  | x |  | *x* | *x* | *x* |  |  |
| 116 | Chronic pain, psychological distress, and quality of life in males with Duchenne muscular dystrophy. | research paper | Huang M, Chen T, Wang Y, Zhou C, Cao J, Lu X, Zeng H. | 2023 | *Developmental Medicine & Child Neurology* | 45 | x |  |  |  |  |  |  | *x* |
| 117 | Behavioural strengths and difficulties in relation to intellectual functions and age in Swedish boys with Duchenne muscular dystrophy | research paper | Gillenstrand J, Ekström AB, Kroksmark AK, Tulinius M, Broberg M. | 2023 | *Child Neuropsychology* | 70 | x |  |  | *x* |  |  |  | *x* |
| 118 | Psychopharmaceutical treatment for neurobehavioral problems in Duchenne muscular dystrophy: a descriptive study using real-world data. | research paper | Weerkamp PMM, Geuens S, Collin P, Goemans N, Vermeulen RJ, De Waele L, Hendriksen JGM, Klinkenberg S. | 2023 | *Neuromuscular disorders* | 52 | x |  |  | *x* |  |  | *x* | *x* |

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| **Supplementary table 2.** DMD behaviors frequently reported by mothers in the 6-11 years old group (n = 24) | | | |
| **CBCL item** | **Behavioral item** | **Frequency** | **Severity** |
| 8 | Can't concentrate, can't pay attention for long | 83% | 1.17 |
| 78 | Inattentive or easily distracted | 79% | 1.17 |
| 19 | Demands a lot of attention | 71% | 1.08 |
| 11 | Clings to adults or too dependent | 67% | 0.83 |
| 1 | Acts too young for his/her age | 63% | 0.71 |
| 17 | Daydreams or gets lost in his/her toughts | 63% | 0.92 |
| 86 | Stobburn, sullen, or irritable | 63% | 0.79 |
| 109 | Whining | 63% | 0.88 |
| 4 | Fails to finish things he/she starts | 58% | 0.63 |
| 22 | Disobedient at home | 58% | 0.58 |
| 45 | Nervous, highstrung, or tense | 54% | 0.58 |
| 62 | Poorly coordinated or clumsy | 54% | 0.63 |
| 95 | Temper tantrums or hot temper | 54% | 0.58 |
| *CBCL = Child Behavior Checklist; Severity scores are reported as means; frequency scores are reported as percentages* | | | |

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| **Supplementary table 3.** DMD behaviors frequently reported by mothers in the 12-18 years old group (n = 24) | | | |
| **CBCL item** | **Behavioral item** | **Frequency** | **Severity** |
| 11 | Clings to adults or too dependent | 79% | 1.29 |
| 1 | Acts too young for his/her age | 71% | 1.04 |
| 19 | Demands a lot of attention | 67% | 0.92 |
| 8 | Can't concentrate, can't pay attention for long | 58% | 0.75 |
| 17 | Daydreams or gets lost in his/her toughts | 58% | 0.71 |
| 102 | Underactive, slow moving, or lacks energy | 58% | 0.83 |
| 9 | Can't get his/her mind off certain thoughts | 54% | 0.63 |
| 42 | Would rather be alone than with others | 54% | 0.63 |
| 86 | Stobburn, sullen, or irritable | 54% | 0.58 |
| 112 | Worries | 54% | 0.67 |
| *CBCL = Child Behavior Checklist; Severity scores are reported as means; frequency scores are reported as percentages* | | | |

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| **Supplementary table 4.** DMD behaviors frequently reported by fathers in the 6-11 years old group (n = 20) | | | |
| **CBCL item** | **Behavioral item** | **Frequency** | **Severity** |
| 8 | Can't concentrate, can't pay attention for long | 95% | 1.32 |
| 11 | Clings to adults or too dependent | 79% | 0.95 |
| 1 | Acts too young for his/her age | 74% | 0.95 |
| 78 | Inattentive or easily distracted | 74% | 0.95 |
| 86 | Stobburn, sullen, or irritable | 74% | 1 |
| 36 | Gets hurt a lot, accident prone | 68% | 0.84 |
| 19 | Demands a lot of attention | 63% | 1 |
| 9 | Can't get his/her mind off certain thougths; obsessions | 58% | 0.84 |
| 109 | Whining | 58% | 0.63 |
| 4 | Fails to finish things he/she started | 53% | 0.63 |
| 74 | Showing off or clowning | 53% | 0.63 |
| *CBCL = Child Behavior Checklist; Severity scores are reported as means; frequency scores are reported as percentages* | | | |

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| **Supplementary table 5.** DMD behaviors frequently reported by fathers in the 12-18 years old group (n = 24) | | | |
| **CBCL item** | **Behavioral item** | **Frequency** | **Severity** |
| 1 | Acts too young for his/her age | 76% | 1.12 |
| 11 | Clings to adults or too dependent | 71% | 1 |
| 19 | Demands a lot of attention | 65% | 0.82 |
| 8 | Can't concentrate, can't pay attention for long | 59% | 0.88 |
| 5 | There is very little he/she enjoys | 53% | 0.59 |
| 75 | Too shy or timid | 53% | 0.71 |
| *CBCL = Child Behavior Checklist; Severity scores are reported as means; frequency scores are reported as percentages* | | | |

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| **Supplementary table 6.** Main results on DuMAND Checklist subscales and instruments used for construct validation | | | | |
|  | **N** | **Mean** | **SD** | **Range** |
| **DuMAND Checklist** |  |  |  |  |
| Cognition & learning | 20 | 21.85 | 7.07 | 9-33 |
| Social responsiveness | 20 | 27.60 | 1.13 | 12-40 |
| Emotion regulation | 20 | 27.85 | 7.81 | 17-43 |
| Externalizing behavior | 20 | 24.70 | 10.28 | 11-41 |
| Eating & sleeping | 20 | 7.85 | 2.56 | 4-13 |
|  |  |  |  |  |
| **CBCL 6 -18 years** |  |  |  |  |
| School | 17 | 35.59 | 9.49 | 20-55 |
|  |  |  |  |  |
| **PARS-III** |  |  |  |  |
| Hostility | 17 | 3.88 | 3.66 | 0-13 |
| Anxiety - depression | 17 | 3.35 | 3.64 | 0-14 |
| Withdrawal | 16 | 1.13 | 1.54 | 0-5 |
|  |  |  |  |  |
| **SRS-2** |  |  |  |  |
| Total scale score | 20 | 63.90 | 14.55 | 37-98 |
|  |  |  |  |  |
| **SDQ** |  |  |  |  |
| Emotional symptoms | 18 | 3.11 | 2.11 | 0-8 |
| Conduct problems | 18 | 1.11 | 1.32 | 0-4 |
| Hyperactivity-Inattention | 18 | 3.56 | 2.60 | 0-8 |
| Externalizing symptoms scale | 17 | 4.76 | 3.63 | 0-12 |
| Internalizing symptoms scale | 17 | 5.06 | 3.11 | 0-9 |
| *DuMAND = Duchenne Muscular dystrophy-Associated Neurobehavioral Difficulties; SD = Standard Deviation; CBCL = Child Behavior Checklist; PARS-III = Personal Adjustment & Role Skills Scale 3rd edition; SRS-2 = Social Responsiveness Scale, 2nd edition; SDQ = Strengths and Difficulties Questionnaire* | | | | |