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The Duchenne brain

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References

A

- Ainslie, Philip N., James D. Cotter, Keith P. George, Sam Lucas, Carissa Murrell, Rob Shave, Kate N. Thomas, Michael J. A. Williams, and Greg Atkinson. 2008. "Elevation in Cerebral Blood Flow Velocity with Aerobic Fitness throughout Healthy Human Ageing." *The Journal of Physiology* 586 (16): 4005–10.
- Almomani, Rowida, Nienke van der Stoep, Egbert Bakker, Johan T den Dunnen, Martijn H Breuning, and Ieke B Ginjaar. 2009. "Rapid and Cost Effective Detection of Small Mutations in the DMD Gene by High Resolution Melting Curve Analysis." *Neuromuscular Disorders : NMD* 19 (6): 383–90.
- Alsop, David C, John A Detre, Xavier Golay, Matthias Günther, Jeroen Hendrikse, Luis Hernandez-Garcia, Hanzhang Lu, et al. 2015. "Recommended Implementation of Arterial Spin-Labeled Perfusion MRI for Clinical Applications: A Consensus of the ISMRM Perfusion Study Group and the European Consortium for ASL in Dementia." *Magnetic Resonance in Medicine* 73 (1): 102–16.
- Amlien, I.K., and A.M. Fjell. 2014. "Diffusion Tensor Imaging of White Matter Degeneration in Alzheimer's Disease and Mild Cognitive Impairment." *Neuroscience* 276 (September): 206–15.
- Asllani, Iris, Ajna Borogovac, and Truman R. Brown. 2008. "Regression Algorithm Correcting for Partial Volume Effects in Arterial Spin Labeling MRI." *Magnetic Resonance in Medicine* 60 (6): 1362–71.
- Attwell, David, Alastair M Buchan, Serge Charpak, Martin Lauritzen, Brian A Macvicar, and Eric A Newman. 2010. "Glial and Neuronal Control of Brain Blood Flow." *Nature* 468 (7321): 232–43.
- Austin, R C, P L Howard, V N D'Souza, H J Klamut, and P N Ray. 1995. "Cloning and Characterization of Alternatively Spliced Isoforms of Dp71." *Hum.Mol.Genet.* 4 (9): 1475–83.
- Assaf, Yaniv, and Ofer Pasternak. 2008. "Diffusion Tensor Imaging (DTI)-Based White Matter Mapping in Brain Research: A Review." *Journal of Molecular Neuroscience : MN* 34 (1): 51–61.

B

- Banihani, R, S Smile, G Yoon, A Dupuis, M Mosleh, A Snider, and L McAdam. 2015. "Cognitive and Neurobehavioral Profile in Boys With Duchenne Muscular Dystrophy." *J.Child Neurol.* 30 (11): 1472–82.
- Bartha, Robert, Shalom Michaeli, Hellmut Merkle, Gregor Adriany, Peter Andersen, Wei Chen, Kamil Ugurbil, and Michael Garwood. 2002. "In Vivo $^1\text{H}_2\text{O}$ T_2 Measurement in the Human Occipital Lobe at 4T and 7T by Carr-Purcell MRI: Detection of Microscopic Susceptibility Contrast." *Magnetic Resonance in Medicine* 47 (4): 742–50.
- Basu, S N, R Kollu, and S Banerjee-Basu. 2009. "AutDB: A Gene Reference Resource for Autism Research." *Nucleic Acids Res.* 37 (Database issue): D832–36.

- Benjamini, Y, and Hochberg Y. 1995. "Controlling the False Discovery Rate - A Practical and Powerful Approach to Multiple Testing." *Journal of the Royal Statistical Society Series B-Methodological*, no. 57: 289–300.
- Billard, C, P Gillet, J L Signoret, E Uicaut, P Bertrand, M Fardeau, M A Barthez-Carpentier, and J J Santini. 1992. "Cognitive Functions in Duchenne Muscular Dystrophy: A Reappraisal and Comparison with Spinal Muscular Atrophy." *Neuromuscul.Disord.* 2 (5–6): 371–78.
- Bladen, Catherine L., David Salgado, Soledad Monges, Maria E. Foncuberta, Kyriaki Kekou, Konstantina Kosma, Hugh Dawkins, et al. 2015. "The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations." *Human Mutation* 36 (4): 395–402.
- Bresolin, N, E Castelli, G P Comi, G Felisari, A Bardoni, D Perani, F Grassi, A Turconi, F Mazzucchelli, and D Gallotti. 1994. "Cognitive Impairment in Duchenne Muscular Dystrophy." *Neuromuscular Disorders : NMD* 4 (4): 359–69.
- Brooke, M H, G M Fenichel, R C Griggs, J R Mendell, R T Moxley, J P Miller, K K Kaiser, J M Florence, S Pandya, and L Signore. 1987. "Clinical Investigation of Duchenne Muscular Dystrophy. Interesting Results in a Trial of Prednisone." *Archives of Neurology* 44 (8): 812–17.
- Brown, E Sherwood, Dixie J Woolston, and Alan B Frol. 2008. "Amygdala Volume in Patients Receiving Chronic Corticosteroid Therapy." *Biological Psychiatry* 63 (7): 705–9.
- Bullmore, E T, J Suckling, S Overmeyer, S Rabe-Hesketh, E Taylor, and M J Brammer. 1999. "Global, Voxel, and Cluster Tests, by Theory and Permutation, for a Difference between Two Groups of Structural MR Images of the Brain." *IEEE Transactions on Medical Imaging* 18 (1): 32–42.
- Byers, T J, H G W Lidov, and L M Kunkel. 1993. "An Alternative Dystrophin Transcript Specific to Peripheral-Nerve." *Nature Genetics* 4 (1): 77–81.
- C**
- Carretta, Donatella, Marialaura Santarelli, Alessandro Sbriccoli, Francesco Pinto, Claudio Catini, and Diego Minciacchi. 2004. "Spatial Analysis Reveals Alterations of Parvalbumin- and Calbindin-Positive Local Circuit Neurons in the Cerebral Cortex of Mutant Mdx Mice." *Brain Research* 1016 (1): 1–11.
- Carretta, Donatella, Marialaura Santarelli, Duccio Vanni, Sonia Ciabatti, Alessandro Sbriccoli, Francesco Pinto, and Diego Minciacchi. 2003. "Cortical and Brainstem Neurons Containing Calcium-Binding Proteins in a Murine Model of Duchenne's Muscular Dystrophy: Selective Changes in the Sensorimotor Cortex." *The Journal of Comparative Neurology* 456 (1): 48–59.
- Castles, a, and M Coltheart. 1993. "Varieties of Developmental Dyslexia." *Cognition* 47 (2): 149–80.
- Chamova, T, V Guerguelcheva, M Raycheva, T Todorov, J Genova, S Bichev, V Bojinova, V Mitev, I Tournev, and A Todorova. 2013. "Association Between Loss of Dp140 and Cognitive Impairment in Duchenne and Becker Dystrophies." *Balkan Journal of Medical Genetics* 16 (1): 21–29.

- Chaussonot, R, J M Edeline, Bec B Le, Massiou N El, S Laroche, and C Vaillend. 2015. "Cognitive Dysfunction in the Dystrophin-Deficient Mouse Model of Duchenne Muscular Dystrophy: A Reappraisal from Sensory to Executive Processes." *Neurobiol.Learn.Mem.* 124: 111–22.
- Chen, J, E E Bardes, B J Aronow, and A G Jegga. 2009. "ToppGene Suite for Gene List Enrichment Analysis and Candidate Gene Prioritization." *Nucleic Acids Res.* 37 (Web Server issue): W305–11.
- Chiron, C, F Pinton, M C Masure, C Duvelleroy-Hommet, F Leon, and C Billard. 1999. "Hemispheric Specialization Using SPECT and Stimulation Tasks in Children with Dysphasia and Dystrophia." *Developmental Medicine and Child Neurology* 41 (8): 512–20.
- Coburn-Litvak, P S, D A Tata, H E Gorby, D P McCloskey, G Richardson, and B J Anderson. 2004. "Chronic Corticosterone Affects Brain Weight, and Mitochondrial, but Not Glial Volume Fraction in Hippocampal Area CA3." *Neuroscience* 124 (2): 429–38.
- Cotton, S, N J Voudouris, and K M Greenwood. 2001. "Intelligence and Duchenne Muscular Dystrophy: Full-Scale, Verbal, and Performance Intelligence Quotients." *Developmental Medicine and Child Neurology* 43 (7): 497–501.
- Cotton, Sue M, Nicholas J Voudouris, and Kenneth M Greenwood. 2005. "Association between Intellectual Functioning and Age in Children and Young Adults with Duchenne Muscular Dystrophy: Further Results from a Meta-Analysis." *Developmental Medicine and Child Neurology* 47 (4): 257–65.
- Courchesne, E, H J Chisum, J Townsend, A Cowles, J Covington, B Egaas, M Harwood, S Hinds, and G A Press. 2000. "Normal Brain Development and Aging: Quantitative Analysis at in Vivo MR Imaging in Healthy Volunteers." *Radiology* 216 (3): 672–82.
- Crosbie, R H. 2001. "NO Vascular Control in Duchenne Muscular Dystrophy." *Nature Medicine* 7 (1): 27–29.
- Cyrułnik, S E, and V J Hinton. 2008. "Duchenne Muscular Dystrophy: A Cerebellar Disorder?" *Neuroscience and Biobehavioral Reviews* 32 (3): 486–96.
- Cyrułnik, Shana E, Robert J Fee, Darryl C De Vivo, Edward Goldstein, and Veronica J Hinton. 2007. "Delayed Developmental Language Milestones in Children with Duchenne's Muscular Dystrophy." *The Journal of Pediatrics* 150 (5): 474–78.
- Cyrułnik, Shana E., Robert J. Fee, Abigail Batchelder, Jacqueline Kiefel, Edward Goldstein, and Veronica J. Hinton. 2008. "Cognitive and Adaptive Deficits in Young Children with Duchenne Muscular Dystrophy (DMD)." *Journal of the International Neuropsychological Society* 14 (5): 853–61.

D

- Davidson, Z E, M M Ryan, A J Kornberg, K Sinclair, A Cairns, K Z Walker, and H Truby. 2014. "Observations of Body Mass Index in Duchenne Muscular Dystrophy: A Longitudinal Study." *European Journal of Clinical Nutrition* 68 (8): 892–97.

- Davies, K E, T J Smith, S Bunday, A P Read, T Flint, M Bell, and A Speer. 1988. "Mild and Severe Muscular Dystrophy Associated with Deletions in Xp21 of the Human X Chromosome." *Journal of Medical Genetics* 25 (1): 9–13.
- de, Ligt J, M H Willemsen, B W van Bon, T Kleefstra, H G Yntema, T Kroes, A T Vulto-van Silfhout, et al. 2012. "Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability." *N.Engl.J.Med.* 367 (20): 1921–29.
- Dooreneer, Nathalie, Eve M. Dumas, Eidrees Ghariq, Sophie Schmid, Chiara S.M. Straathof, Arno A.W. Roest, Beatrijs H. Wokke, et al. 2016. "Decreased Cerebral Perfusion in Duchenne Muscular Dystrophy Patients." *Neuromuscular Disorders*.
- Dooreneer, N., C.S. Straathof, E.M. Dumas, P. Spitali, I.B. Ginjaar, B.H. Wokke, D.G. Schrans, et al. 2014. "Reduced Cerebral Gray Matter and Altered White Matter in Boys with Duchenne Muscular Dystrophy." *Annals of Neurology* 76 (3): 403–11.
- Dorman, C, A D Hurley, and J D'Avignon. 1988. "Language and Learning Disorders of Older Boys with Duchenne Muscular Dystrophy." *Dev.Med.Child Neurol.* 30 (3): 316–27.
- Dougherty, Chase C., David W. Evans, Scott M. Myers, Gregory J. Moore, and Andrew M. Michael. 2016. "A Comparison of Structural Brain Imaging Findings in Autism Spectrum Disorder and Attention-Deficit Hyperactivity Disorder." *Neuropsychology Review* 26 (1): 25–43.
- Dsouza, V N, N T Man, G E Morris, W Karges, D A M Pillers, and P N Ray. 1995. "A Novel Dystrophin Isoform Is Required for Normal Retinal Electrophysiology." *Human Molecular Genetics* 4 (5): 837–42.
- Dubowitz, V, and L Crome. 1969. "The Central Nervous System in Duchenne Muscular Dystrophy." *Brain : A Journal of Neurology* 92 (4): 805–8.
- E**
- Emery, Alan E. H., Francesco Muntoni, and Rosaline C. M. Quinlivan. 2015. *Duchenne Muscular Dystrophy*. Oxford University Press.
- Emery, Alan E H, E Meryon, E Meryon, AEH Emery, MLH Emery, GBA Duchenne, GBA Duchenne, et al. 2002. "The Muscular Dystrophies." *Lancet (London, England)* 359 (9307). Elsevier: 687–95.
- Emmer, Bart J, Matthias J van Osch, Ona Wu, Gerda M Steup-Beekman, Stefan C Steens, Tom W Huizinga, Mark A van Buchem, and Jeroen van der Grond. 2010. "Perfusion MRI in Neuro-Psychiatric Systemic Lupus Erythematosus." *Journal of Magnetic Resonance Imaging : JMRI* 32 (2): 283–88.
- Ernst T, Kreis R, Ross BD. Absolute Quantitation of Water and Metabolites in the Human Brain. I. Compartments and Water. *J. Magn. Reson. Ser. B* 1993; 102: 1–8.

F

- Fatemi, S Hossein, Kimberly A Aldinger, Paul Ashwood, Margaret L Bauman, Charles D Blaha, Gene J Blatt, Abha Chauhan, et al. 2012. "Consensus Paper: Pathological Role of the Cerebellum in Autism." *Cerebellum* (London, England) 11 (3). NIH Public Access: 777–807.
- Feener, Chris Anne, Michel Koenig, and Louis M. Kunkel. 1989. "Alternative Splicing of Human Dystrophin mRNA Generates Isoforms at the Carboxy Terminus." *Nature* 338 (6215): 509–11.
- Felisari, G, F Martinelli Boneschi, A Bardoni, M Sironi, G P Comi, M Robotti, A C Turconi, M Lai, G Corrao, and N Bresolin. 2000. "Loss of Dp140 Dystrophin Isoform and Intellectual Impairment in Duchenne Dystrophy." *Neurology* 55 (4): 559–64.
- Forrest, A R, H Kawaji, M Rehli, J K Baillie, M J de Hoon, V Haberle, T Lassmann, et al. 2014. "A Promoter-Level Mammalian Expression Atlas." *Nature* 507 (7493): 462–70.

G

- Gasparovic, Charles, Tao Song, Deidre Devier, H. Jeremy Bockholt, Arvind Caprihan, Paul G. Mullins, Stefan Posse, Rex E. Jung, and Leslie A. Morrison. 2006. "Use of Tissue Water as a Concentration Reference for Proton Spectroscopic Imaging." *Magnetic Resonance in Medicine* 55 (6): 1219–26.
- Good, Catriona D., Ingrid S. Johnsrude, John Ashburner, Richard N.A. Henson, Karl J. Friston, and Richard S.J. Frackowiak. 2001. "A Voxel-Based Morphometric Study of Ageing in 465 Normal Adult Human Brains." *NeuroImage* 14 (1): 21–36.
- Goodman, A., and R. Goodman. 2012. "Strengths and Difficulties Questionnaire Scores and Mental Health in Looked after Children." *The British Journal of Psychiatry* 200 (5): 426–27.
- Goodnough, Candida L, Ying Gao, Xin Li, Mohammed Q Qutaish, L Henry Goodnough, Joseph Molter, David Wilson, Chris A Flask, and Xin Yu. 2014. "Lack of Dystrophin Results in Abnormal Cerebral Diffusion and Perfusion in Vivo." *NeuroImage* 102 Pt 2 (0 2). NIH Public Access: 809–16.
- Goodwin, F, F Muntoni, and V Dubowitz. 1997. "Epilepsy in Duchenne and Becker Muscular Dystrophies." *Eur.J.Paediatr.Neurol.* 1 (4): 115–19.
- Gorecki, D C, A P Monaco, J M J Derry, A P Walker, E A Barnard, and P J Barnard. 1992. "Expression of 4 Alternative Dystrophin Transcripts in Brain-Regions Regulated by Different Promoters." *Human Molecular Genetics* 1 (7): 505–10.
- Goyenvalle, Aurélie, Graziella Griffith, Arran Babbs, Samir El Andaloussi, Kariem Ezzat, Aurélie Avril, Branislav Dugovic, et al. 2015. "Functional Correction in Mouse Models of Muscular Dystrophy Using Exon-Skipping Tricyclo-DNA Oligomers." *Nature Medicine* 21 (3): 270–75.
- Griffa, Alessandra, Philipp S. Baumann, Jean-Philippe Thiran, and Patric Hagmann. 2013. "Structural Connectomics in Brain Diseases." *NeuroImage* 80 (October): 515–26.

- Griffin, J L, H J Williams, E Sang, K Clarke, C Rae, and J K Nicholson. 2006. "Metabolic Profiling of Genetic Disorders : A Multitissue ¹H Nuclear Magnetic Resonance Spectroscopic and Pattern Recognition Study into Dystrophic Tissue." *Analytical Biochemistry* 21 (2001): 16–21.
- Gruhn, N, F S Larsen, S Boesgaard, G M Knudsen, S A Mortensen, G Thomsen, and J Aldershvile. 2001. "Cerebral Blood Flow in Patients with Chronic Heart Failure before and after Heart Transplantation." *Stroke* 32 (11): 2530–33.
- H**
- Hales, Patrick W, Jamie M Kawadler, Sarah E Aylett, Fenella J Kirkham, and Christopher A Clark. 2014. "Arterial Spin Labeling Characterization of Cerebral Perfusion during Normal Maturation from Late Childhood into Adulthood: Normal 'Reference Range' Values and Their Use in Clinical Studies." *Journal of Cerebral Blood Flow and Metabolism : Official Journal of the International Society of Cerebral Blood Flow and Metabolism* 34 (5): 776–84.
- Hall, Catherine N, Clare Reynell, Bodil Gesslein, Nicola B Hamilton, Anusha Mishra, Brad A Sutherland, Fergus M O'Farrell, Alastair M Buchan, Martin Lauritzen, and David Attwell. 2014. "Capillary Pericytes Regulate Cerebral Blood Flow in Health and Disease." *Nature* 508 (7494): 55–60.
- Hawrylycz, M J, E S Lein, A L Guillozet-Bongaarts, E H Shen, L Ng, J A Miller, L N van de Lagemaat, et al. 2012. "An Anatomically Comprehensive Atlas of the Adult Human Brain Transcriptome." *Nature* 489 (7416): 391–99.
- Hendriksen, J G, and J S Vles. 2008. "Neuropsychiatric Disorders in Males with Duchenne Muscular Dystrophy: Frequency Rate of Attention-Deficit Hyperactivity Disorder (ADHD), Autism Spectrum Disorder, and Obsessive--Compulsive Disorder." *J.Child Neurol.* 23 (5): 477–81.
- Hendriksen, Jos G M, and Johan S H Vles. 2006. "Are Males with Duchenne Muscular Dystrophy at Risk for Reading Disabilities?" *Pediatric Neurology* 34 (4): 296–300.
- Hendriksen, Jos G M, James T Poysky, Debby G M Schrans, Eric G W Schouten, Albert P Aldenkamp, and Johan S H Vles. 2007. "Psychosocial Adjustment in Males with Duchenne Muscular Dystrophy: Psychometric Properties and Clinical Utility of a Parent-Report Questionnaire." *Journal of Pediatric Psychology* 34 (1): 69–78.
- Hendriksen, Ruben G F, Sandra Schipper, Govert Hoogland, Olaf E M G Schijns, Jim T A Dings, Marlien W Aalbers, and Johan S H Vles. 2016. "Dystrophin Distribution and Expression in Human and Experimental Temporal Lobe Epilepsy." *Frontiers in Cellular Neuroscience* 10. *Frontiers Media SA*: 174.
- Hinton, V J, D C De Vivo, N E Nereo, E Goldstein, and Y Stern. 2000. "Poor Verbal Working Memory across Intellectual Level in Boys with Duchenne Dystrophy." *Neurology* 54 (11): 2127–32.
- Holder, E, M Maeda, and R D Bies. 1996. "Expression and Regulation of the Dystrophin Purkinje Promoter in Human Skeletal Muscle, Heart, and Brain." *Human Genetics* 97 (2): 232–39.

Hormozdiari, F, O Penn, E Borenstein, and E E Eichler. 2015. "The Discovery of Integrated Gene Networks for Autism and Related Disorders." *Genome Res.* 25 (1): 142–54.

I

Iossifov, I, M Ronemus, D Levy, Z Wang, I Hakker, J Rosenbaum, B Yamrom, et al. 2012. "De Novo Gene Disruptions in Children on the Autistic Spectrum." *Neuron* 74 (2): 285–99.

J

Jagadha, V, and L E Becker. 2017. "Brain Morphology in Duchenne Muscular Dystrophy: A Golgi Study." *Pediatric Neurology* 4 (2): 87–92. Accessed April 20.

Judge, Daniel P., David A. Kass, W. Reid Thompson, and Kathryn R. Wagner. 2011. "Pathophysiology and Therapy of Cardiac Dysfunction in Duchenne Muscular Dystrophy." *American Journal Cardiovascular Drugs* 11 (5): 287–94.

K

Kato, Toshinori, C A Masami Nishina, Kazuhiro Matsushita, Eitaro Hori, Shinjiro Akaboshi, and Sachio Takashima. 1997. "Increased Cerebral Choline-Compounds in Duchenne Muscular Dystrophy." *Clinical Neuroscience and Neuropsychology* 8 (6): 1435–37.

Kilroy, Emily, Collin Y Liu, Lirong Yan, Yoon Chun Kim, Mirella Dapretto, Mario F Mendez, and Danny J J Wang. 2011. "Relationships between Cerebral Blood Flow and IQ in Typically Developing Children and Adolescents." *Journal of Cognitive Science* 12 (2): 151–70.

Kinnett, Kathi, Sunil Rodger, Elizabeth Vroom, Pat Furlong, Annemieke Aartsma-Rus, and Kate Bushby. 2015. "Imperatives for DUCHENNE MD: A Simplified Guide to Comprehensive Care for Duchenne Muscular Dystrophy." *PLoS Currents*. Public Library of Science.

Koenig, M, A H Beggs, M Moyer, S Scherpf, K Heindrich, T Bettecken, G Meng, et al. 1989. "The Molecular Basis for Duchenne versus Becker Muscular Dystrophy: Correlation of Severity with Type of Deletion." *American Journal of Human Genetics* 45 (4). Elsevier: 498–506.

Koenig, M., E.P. Hoffman, C.J. Bertelson, A.P. Monaco, C. Feener, and L.M. Kunkel. 1987. "Complete Cloning of the Duchenne Muscular Dystrophy (DMD) cDNA and Preliminary Genomic Organization of the DMD Gene in Normal and Affected Individuals." *Cell* 50 (3): 509–17.

Kreis, Roland, Kevin Wingeier, Peter Vermathen, Elisabeth Giger, Franziska Joncourt, Karin Zwygart, Franz Kaufmann, Chris Boesch, and Maja Steinlin. 2011. "Brain Metabolite Composition in Relation to Cognitive Function and Dystrophin Mutations in Boys with Duchenne Muscular Dystrophy." *NMR in Biomedicine* 24 (3): 253–62.

- Krumm, Niklas, Brian J O’Roak, Jay Shendure, and Evan E Eichler. 2014. “A de Novo Convergence of Autism Genetics and Molecular Neuroscience.” *Trends in Neurosciences* 37 (2). Howard Hughes Medical Institute: 95–105.
- Kueh, S L L, S I Head, and J W Morley. 2008. “GABA(A) Receptor Expression and Inhibitory Post-Synaptic Currents in Cerebellar Purkinje Cells in Dystrophin-Deficient Mdx Mice.” *Clinical and Experimental Pharmacology & Physiology* 35 (2): 207–10.
- Kundaje, A, W Meuleman, J Ernst, M Bilenky, A Yen, A Heravi-Moussavi, P Kheradpour, et al. 2015. “Integrative Analysis of 111 Reference Human Epigenomes.” *Nature* 518 (7539): 317–30.
- L**
- Lebel, C., L. Walker, A. Leemans, L. Phillips, and C. Beaulieu. 2008. “Microstructural Maturation of the Human Brain from Childhood to Adulthood.” *NeuroImage* 40 (3): 1044–55.
- Lederfein, D, Z Levy, N Augier, D Mornet, G Morris, O Fuchs, D Yaffe, and U Nudel. 1992. “A 71-Kilodalton Protein Is a Major Product of the Duchenne Muscular Dystrophy Gene in Brain and Other Nonmuscle Tissues.” *Proc.Natl.Acad.Sci.U.S.A* 89 (12): 5346–50.
- Lee, Joon Soo, Zoltán Pfünd, Csaba Juhász, Michael E Behen, Otto Muzik, Diane C Chugani, Michael a Nigro, and Harry T Chugani. 2002. “Altered Regional Brain Glucose Metabolism in Duchenne Muscular Dystrophy: A Pet Study.” *Muscle & Nerve* 26 (4): 506–12.
- Lee, Tomoko, Yasuhiro Takeshima, Noriko Kusunoki, Hiroyuki Awano, Mariko Yagi, Masafumi Matsuo, and Kazumoto Iijima. 2014. “Differences in Carrier Frequency between Mothers of Duchenne and Becker Muscular Dystrophy Patients.” *Journal of Human Genetics* 59 (1). Nature Publishing Group: 46–50.
- Leemans, A, B Jeurissen, and Sijbers J. 2009. “No Title.” In *ExploreDTI: A Graphical Toolbox for Processing, Analysing, and Visualizing Diffusion MR Data*. 17th Annual Meeting of Intl Soc Mag Reson Med.
- Lenhard, B, A Sandelin, and P Carninci. 2012. “Metazoan Promoters: Emerging Characteristics and Insights into Transcriptional Regulation.” *Nat.Rev.Genet.* 13 (4): 233–45.
- Lidov, H G, S Selig, and L M Kunkel. 1995. “Dp140: A Novel 140 kDa CNS Transcript from the Dystrophin Locus.” *Human Molecular Genetics* 4 (3): 329–35.
- Lidov, H G W, T J Byers, and L M Kunkel. 1993. “The Distribution of Dystrophin in the Murine Central-Nervous-System - An Immunocytochemical Study.” *Neuroscience* 54 (1): 167–87.
- Lidov, H G W. 1996. “Dystrophin in the Nervous System.” *Brain Pathology* 6 (1): 63–77.
- Lidov, H G W, T J Byers, S C Watkins, and L M Kunkel. 1990. “Localization of Dystrophin to Postsynaptic Regions of Central-Nervous-System Cortical-Neurons.” *Nature* 348 (6303): 725–27.

- Lim, Kenji Rowel, Rika Maruyama, and Toshifumi Yokota. 2017. "Eteplirsen in the Treatment of Duchenne Muscular Dystrophy." *Drug Design, Development and Therapy* Volume 11 (February): 533–45.
- Lindberg, Ulrich, Nanna Witting, Stine Lundgaard Jørgensen, John Vissing, Egill Rostrup, Henrik Bo Wiberg Larsson, and Christina Kruuse. 2017. "Effects of Sildenafil on Cerebrovascular Reactivity in Patients with Becker Muscular Dystrophy." *Neurotherapeutics : The Journal of the American Society for Experimental NeuroTherapeutics* 14 (1): 182–90.
- Liu, Yansheng, Andreas Beyer, Ruedi Aebersold, J.S. Parker, D.N. Hayes, C.M. Perou, M.C. Chambers, et al. 2016. "On the Dependency of Cellular Protein Levels on mRNA Abundance." *Cell* 165 (3). The Royal Society of Chemistry: 535–50.
- Lizio, M, J Harshbarger, H Shimoji, J Severin, T Kasukawa, S Sahin, I Abugessaisa, et al. 2015. "Gateways to the FANTOM5 Promoter Level Mammalian Expression Atlas." *Genome.Biol.* 16: 22.
- Loufrani, L., Caroline Dubroca, Dong You, Z Li, Bernard Levy, Denise Paulin, and Daniel Henrion. 2004. "Absence of Dystrophin in Mice Reduces NO-Dependent Vascular Function and Vascular Density: Total Recovery After a Treatment with the Aminoglycoside Gentamicin." *Arteriosclerosis, Thrombosis, and Vascular Biology* 24 (4): 671–76.
- Lv, S-Y, Q-H Zou, J-L Cui, N Zhao, J Hu, X-Y Long, Y-C Sun, et al. 2011. "Decreased Gray Matter Concentration and Local Synchronization of Spontaneous Activity in the Motor Cortex in Duchenne Muscular Dystrophy." *AJNR. American Journal of Neuroradiology*, September, 1–5.

M

- Mah, Jean. 2016. "Current and Emerging Treatment Strategies for Duchenne Muscular Dystrophy." *Neuropsychiatric Disease and Treatment* Volume 12 (July): 1795–1807.
- Mah, Jean K., Lawrence Korngut, Jonathan Dykeman, Lundy Day, Tamara Pringsheim, and Nathalie Jette. 2014. "A Systematic Review and Meta-Analysis on the Epidemiology of Duchenne and Becker Muscular Dystrophy." *Neuromuscular Disorders* 24 (6): 482–91.
- Matthews, Emma, Ruth Brassington, Thierry Kuntzer, Fatima Jichi, and Adnan Y Manzur. 2016. "Corticosteroids for the Treatment of Duchenne Muscular Dystrophy." In *Cochrane Database of Systematic Reviews*, edited by Emma Matthews, CD003725. Chichester, UK: John Wiley & Sons, Ltd.
- Marjańska, Małgorzata, Edward J Auerbach, Romain Valabrègue, Pierre-François Van de Moortele, Gregor Adriany, and Michael Garwood. 2012. "Localized 1H NMR Spectroscopy in Different Regions of Human Brain in Vivo at 7T: T2 Relaxation Times and Concentrations of Cerebral Metabolites." *NMR in Biomedicine* 25 (2). NIH Public Access: 332–39.
- Mendell, Jerry R., Louise R. Rodino-Klapac, Zarife Sahenk, Kandice Roush, Loren Bird, Linda P. Lowes, Lindsay Alfano, et al. 2013. "Eteplirsen for the Treatment of Duchenne Muscular Dystrophy." *Annals of Neurology* 74 (5): 637–47.

- Mensen, Vincent T, Lara M Wierenga, Sarai van Dijk, Yvonne Rijks, Bob Oranje, René C W Mandl, and Sarah Durston. 2017. "Development of Cortical Thickness and Surface Area in Autism Spectrum Disorder." *NeuroImage. Clinical* 13: 215–22.
- Miller, J A, S L Ding, S M Sunkin, K A Smith, L Ng, A Szafer, A Ebbert, et al. 2014. "Transcriptional Landscape of the Prenatal Human Brain." *Nature* 508 (7495): 199–206.
- Moizard, M P, a Toutain, D Fournier, F Berret, M Raynaud, C Billard, C Andres, and C Moraine. 2000. "Severe Cognitive Impairment in DMD: Obvious Clinical Indication for Dp71 Isoform Point Mutation Screening." *European Journal of Human Genetics : EJHG* 8 (7): 552–56.
- Monaco, Anthony P., Rachael L. Neve, Chris Colletti-Feener, Corlee J. Bertelson, David M. Kurnit, and Louis M. Kunkel. 1986. "Isolation of Candidate cDNAs for Portions of the Duchenne Muscular Dystrophy Gene." *Nature* 323 (6089): 646–50.
- Morris, G E, C Simmons, and N T Man. 1995. "Apo-Dystrophins (Dp140 and Dp71) and Dystrophin Splicing Isoforms in Developing Brain." *Biochemical and Biophysical Research Communications* 215 (1): 361–67.
- Moser, H. 1984. "Duchenne Muscular Dystrophy: Pathogenetic Aspects and Genetic Prevention." *Human Genetics* 66 (1): 17–40.
- N**
- Neale, B M, Y Kou, L Liu, A Ma'ayan, K E Samocha, A Sabo, C F Lin, et al. 2012. "Patterns and Rates of Exonic de Novo Mutations in Autism Spectrum Disorders." *Nature* 485 (7397): 242–45.
- Nico, Beatrice, Luisa Roncali, Domenica Mangieri, and Domenico Ribatti. 2005. "Blood-Brain Barrier Alterations in MDX Mouse, an Animal Model of the Duchenne Muscular Dystrophy." *Current Neurovascular Research* 2 (1): 47–54.
- Nico, Beatrice, Roberto Tamma, Tiziana Annese, Domenica Mangieri, Annamaria De Luca, Patrizia Corsi, Vincenzo Benagiano, et al. 2010a. "Glial Dystrophin-Associated Proteins, Laminin and Agrin, Are Downregulated in the Brain of Mdx Mouse." *Laboratory Investigation* 90 (11): 1645–60.
- Nudel, U, D Zuk, P Einat, E Zeelon, Z Levy, S Neuman, and D Yaffe. 1989. "Duchenne Muscular-Dystrophy Gene-Product Is Not Identical in Muscle and Brain." *Nature* 337 (6202): 76–78.
- O**
- O'Roak, B J, L Vives, S Girirajan, E Karakoc, N Krumm, B P Coe, R Levy, et al. 2012. "Sporadic Autism Exomes Reveal a Highly Interconnected Protein Network of de Novo Mutations." *Nature* 485 (7397): 246–50.

P

- Pane, M, S Messina, C Bruno, A D'Amico, M Villanova, B Brancalion, S Sivo, et al. 2013. "Duchenne Muscular Dystrophy and Epilepsy." *Neuromuscul.Disord.* 23 (4): 313–15.
- Pane, M, R Scalise, A Berardinelli, G D'Angelo, V Ricotti, P Alfieri, I Moroni, et al. 2013. "Early Neurodevelopmental Assessment in Duchenne Muscular Dystrophy." *Neuromuscul.Disord.* 23 (6): 451–55. doi:S0960-8966(13)00085-0
- Penny, W. 2006. *Statistical Parametric Mapping*. Elsevier.
- Pinero, J, N Queralt-Rosinach, A Bravo, J Deu-Pons, A Bauer-Mehren, M Baron, F Sanz, and L I Furlong. 2015. "DisGeNET: A Discovery Platform for the Dynamical Exploration of Human Diseases and Their Genes." *Database.(Oxford)* 2015: bav028. doi:bav028
- Poels, Mariëlle MF, Mohammad Arfan Ikram, Meike W Vernooij, Gabriel P Krestin, Albert Hofman, Wiro J Messen, Aad van der Lugt, and Monique MB Breteler. 2008. "Total Cerebral Blood Flow in Relation to Cognitive Function: The Rotterdam Scan Study." *Journal of Cerebral Blood Flow & Metabolism* 28 (10): 1652–55.
- Provencher, Stephen W. 2001. "Automatic Quantitation of Localized in Vivo 1H Spectra with LCMoDel." *NMR in Biomedicine* 14 (4): 260–64.

Q

R

- Rae, C, R B Scott, C H Thompson, R M Dixon, I Dumughn, G J Kemp, a Male, M Pike, P Styles, and G K Radda. 1998. "Brain Biochemistry in Duchenne Muscular Dystrophy: A 1H Magnetic Resonance and Neuropsychological Study." *Journal of the Neurological Sciences* 160 (2): 148–57.
- Rauch, A, D Wiczorek, E Graf, T Wieland, S Endeke, T Schwarzmayr, B Albrecht, et al. 2012. "Range of Genetic Mutations Associated with Severe Non-Syndromic Sporadic Intellectual Disability: An Exome Sequencing Study." *Lancet* 380 (9854): 1674–82.
- Reiss, A L, M T Abrams, H S Singer, J L Ross, and M B Denckla. 1996. "Brain Development, Gender and IQ in Children. A Volumetric Imaging Study." *Brain : A Journal of Neurology*, October, 1763–74.
- Ricotti, V, H Jagle, M Theodorou, A T Moore, F Muntoni, and D A Thompson. 2015. "Ocular and Neurodevelopmental Features of Duchenne Muscular Dystrophy: A Signature of Dystrophin Function in the Central Nervous System." *Eur.J.Hum.Genet.*
- Ricotti, V, W P Mandy, M Scoto, M Pane, N Deconinck, S Messina, E Mercuri, D H Skuse, and F Muntoni. 2015. "Neurodevelopmental, Emotional, and Behavioural Problems in Duchenne Muscular Dystrophy in Relation to Underlying Dystrophin Gene Mutations." *Dev.Med.Child Neurol.*

- Robinson-Hamm, Jacqueline N., and Charles A. Gersbach. 2016. "Gene Therapies That Restore Dystrophin Expression for the Treatment of Duchenne Muscular Dystrophy." *Human Genetics* 135 (9): 1029–40.
- Rooney, William D., Glyn Johnson, Xin Li, Eric R. Cohen, Seong Gi Kim, Kamil Ugurbil, and Charles S. Springer. 2007. "Magnetic Field and Tissue Dependencies of Human Brain Longitudinal $1H_2O$ Relaxation in Vivo." *Magnetic Resonance in Medicine* 57 (2): 308–18.
- Ruijter, J M, C Ramakers, W M H Hoogaars, Y Karlen, O Bakker, M J B van den Hoff, and A F M Moorman. 2009. "Amplification Efficiency: Linking Baseline and Bias in the Analysis of Quantitative PCR Data." *Nucleic Acids Research* 37 (6): e45..
- S**
- Sander, M, B Chavoshan, S A Harris, S T Iannaccone, J T Stull, G D Thomas, and R G Victor. 2000. "Functional Muscle Ischemia in Neuronal Nitric Oxide Synthase-Deficient Skeletal Muscle of Children with Duchenne Muscular Dystrophy." *Proceedings of the National Academy of Sciences of the United States of America* 97 (25): 13818–23.
- Sanders, S J, M T Murtha, A R Gupta, J D Murdoch, M J Raubeson, A J Willsey, A G Ercan-Sencicek, et al. 2012. "De Novo Mutations Revealed by Whole-Exome Sequencing Are Strongly Associated with Autism." *Nature* 485 (7397): 237–41.
- Sarrazin, Elisabeth, Maja von der Hagen, Ulrike Schara, Katja von Au, and Angela M Kaindl. 2014. "Growth and Psychomotor Development of Patients with Duchenne Muscular Dystrophy." *European Journal of Paediatric Neurology : EJPN : Official Journal of the European Paediatric Neurology Society* 18 (1): 38–44.
- Sbriccoli, A, M Santarelli, D Carretta, F Pinto, A Granato, and D Minciocchi. 1995. "Architectural Changes of the Cortico-Spinal System in the Dystrophin Defective Mdx Mouse." *Neuroscience Letters* 200 (1): 53–56.
- Sekiguchi, Masayuki, Ko Zushida, Mikiharu Yoshida, Motoko Maekawa, Sari Kamichi, Mizuko Yoshida, Yoshinori Sahara, Shigeki Yuasa, Shin'ichi Takeda, and Keiji Wada. 2009. "A Deficit of Brain Dystrophin Impairs Specific Amygdala GABAergic Transmission and Enhances Defensive Behaviour in Mice." *Brain : A Journal of Neurology* 132 (Pt 1): 124–35.
- Smith, Stephen M. 2002. "Fast Robust Automated Brain Extraction." *Human Brain Mapping* 17 (3): 143–55.
- Smith, Stephen M., Mark Jenkinson, Mark W. Woolrich, Christian F. Beckmann, Timothy E.J. Behrens, Heidi Johansen-Berg, Peter R. Bannister, et al. 2004. "Advances in Functional and Structural MR Image Analysis and Implementation as FSL." *NeuroImage* 23 (January): S208–19.
- Smith, Stephen M, Mark Jenkinson, Heidi Johansen-Berg, Daniel Rueckert, Thomas E Nichols, Clare E Mackay, Kate E Watkins, et al. 2006. "Tract-Based Spatial Statistics: Voxelwise Analysis of Multi-Subject Diffusion Data." *NeuroImage* 31 (4): 1487–1505.

- Snow, Wanda M., Judy E. Anderson, and Lorna S. Jakobson. 2013. "Neuropsychological and Neurobehavioral Functioning in Duchenne Muscular Dystrophy: A Review." *Neuroscience & Biobehavioral Reviews* 37 (5): 743–52.
- Snow, W M, J E Anderson, and M Fry. 2014. "Regional and Genotypic Differences in Intrinsic Electrophysiological Properties of Cerebellar Purkinje Neurons from Wild-Type and Dystrophin-Deficient Mdx Mice." *Neurobiol.Learn.Mem.* 107: 19–31.
- Snow, W M, M Fry, and J E Anderson. 2013. "Increased Density of Dystrophin Protein in the Lateral versus the Vermal Mouse Cerebellum." *Cell Mol.Neurobiol.* 33 (4): 513–20.
- Song, Sheng-Kwei, Jun Yoshino, Tuan Q. Le, Shioh-Juan Lin, Shu-Wei Sun, Anne H. Cross, and Regina C. Armstrong. 2005. "Demyelination Increases Radial Diffusivity in Corpus Callosum of Mouse Brain." *NeuroImage* 26 (1): 132–40.
- Strehle, Eugen-Matthias, and Volker Straub. 2015. "Recent Advances in the Management of Duchenne Muscular Dystrophy." *Archives of Disease in Childhood* 100 (12). BMJ Publishing Group Ltd and Royal College of Paediatrics and Child Health: 1173–77.
- Stuart, J M, E Segal, D Koller, and S K Kim. 2003. "A Gene-Coexpression Network for Global Discovery of Conserved Genetic Modules." *Science* 302 (5643): 249–55.
- Sweeney, H L, and E R Barton. 2000. "The Dystrophin-Associated Glycoprotein Complex: What Parts Can You Do Without?" *Proc.Natl.Acad.Sci.U.S.A.* 97 (25): 13464–66.
- Syed, Yahya Y. 2016. "Eteplirsen: First Global Approval." *Drugs* 76 (17): 1699–1704.
- T**
- Takahashi, T, R Shirane, S Sato, and T Yoshimoto. 1999. "Developmental Changes of Cerebral Blood Flow and Oxygen Metabolism in Children." *AJNR. American Journal of Neuroradiology* 20 (5): 917–22.
- Taki, Yasuyuki, Hiroshi Hashizume, Yuko Sassa, Hikaru Takeuchi, Kai Wu, Michiko Asano, Kohei Asano, Hiroshi Fukuda, and Ryuta Kawashima. 2011. "Correlation between Gray Matter Density-Adjusted Brain Perfusion and Age Using Brain MR Images of 202 Healthy Children." *Human Brain Mapping* 32 (11): 1973–85.
- Taylor, P J, G A Betts, S Maroulis, C Gilissen, R L Pedersen, D R Mowat, H M Johnston, and M F Buckley. 2010. "Dystrophin Gene Mutation Location and the Risk of Cognitive Impairment in Duchenne Muscular Dystrophy." *Plos One* 5 (1).
- Teeuwisse, W. M., W. M. Brink, K. N. Haines, and A. G. Webb. 2012. "Simulations of High Permittivity Materials for 7 T Neuroimaging and Evaluation of a New Barium Titanate-Based Dielectric." *Magnetic Resonance in Medicine* 67 (4). Wiley Subscription Services, Inc., A Wiley Company: 912–18.

Thompson, Paul M., Jason L. Stein, Sarah E. Medland, Derrek P. Hibar, Alejandro Arias Vasquez, Miguel E. Renteria, Roberto Toro, et al. 2014. "The ENIGMA Consortium: Large-Scale Collaborative Analyses of Neuroimaging and Genetic Data." *Brain Imaging and Behavior* 8 (2): 153–82.

Tkáč, I, and R Gruetter. 2005. "Methodology of 1 H NMR Spectroscopy of the Human Brain at Very High Magnetic Fields." *Applied Magnetic Resonance* 29 (1): 139–57.

Torelli, S., S. C. Brown, C. Jimenez-Mallebrera, L. Feng, F. Muntoni, and C. A. Sewry. 2004. "Absence of Neuronal Nitric Oxide Synthase (nNOS) as a Pathological Marker for the Diagnosis of Becker Muscular Dystrophy with Rod Domain Deletions." *Neuropathology and Applied Neurobiology* 30 (5): 540–45.

Townsend, D. 2014. "Finding the Sweet Spot: Assembly and Glycosylation of the Dystrophin-Associated Glycoprotein Complex." *Anat.Rec.(Hoboken.)* 297 (9): 1694–1705.

Tracey, I, J F Dunn, H G Parkes, and G K Radda. 1996. "An in Vivo and in Vitro H-Magnetic Resonance Spectroscopy Study of Mdx Mouse Brain: Abnormal Development or Neural Necrosis?" *Journal of the Neurological Sciences* 141 (1–2): 13–18.

U

V

Vaillend, C, J M Billard, and S Laroche. 2004. "Impaired Long-Term Spatial and Recognition Memory and Enhanced CA1 Hippocampal LTP in the Dystrophin-Deficient Dmd(mdx) Mouse." *Neurobiol.Dis.* 17 (1): 10–20.

van den Bergen, J.C., H.B. Ginjaar, A.J. van Essen, R. Pangalila, I.J.M. de Groot, P.J. Wijkstra, M.P. Zijnen, et al. 2014. "Forty-Five Years of Duchenne Muscular Dystrophy in The Netherlands." *Journal of Neuromuscular Diseases* 1 (1). IOS Press: 99–109.

Vandermosten, Maaïke, Bart Boets, Jan Wouters, and Pol Ghesquière. 2012. "A Qualitative and Quantitative Review of Diffusion Tensor Imaging Studies in Reading and Dyslexia." *Neuroscience & Biobehavioral Reviews* 36 (6): 1532–52.

Verhaert, David, Kathryn Richards, Jill A Rafael-Fortney, and Subha V Raman. 2011. "Cardiac Involvement in Patients with Muscular Dystrophies: Magnetic Resonance Imaging Phenotype and Genotypic Considerations." *Circulation. Cardiovascular Imaging* 4 (1): 67–76.

W

Waite, A, S C Brown, and D J Blake. 2012. "The Dystrophin-Glycoprotein Complex in Brain Development and Disease." *Trends Neurosci.* 35 (8): 487–96.

- Wallace, Gregory L., Ian W. Eisenberg, Briana Robustelli, Nathan Dankner, Lauren Kenworthy, Jay N. Giedd, and Alex Martin. 2015. "Longitudinal Cortical Development During Adolescence and Young Adulthood in Autism Spectrum Disorder: Increased Cortical Thinning but Comparable Surface Area Changes." *Journal of the American Academy of Child & Adolescent Psychiatry* 54 (6): 464–69.
- Wang, Jing-Zhang, Peng Wu, Zhi-Min Shi, Yan-Li Xu, and Zhi-Jun Liu. 2017. "The AAV-Mediated and RNA-Guided CRISPR/Cas9 System for Gene Therapy of DMD and BMD." *Brain and Development*, April.
- Wheway, J M, and R G Roberts. 2003. "The Dystrophin Lymphocyte Promoter Revisited: 4.5-Megabase Intron, or Artifact?" *Neuromuscul.Disord.* 13 (1): 17–20.
- Willeumier, Kristen C., Derek V. Taylor, and Daniel G. Amen. 2011. "Elevated BMI Is Associated With Decreased Blood Flow in the Prefrontal Cortex Using SPECT Imaging in Healthy Adults." *Obesity* 19 (5): 1095–97.
- Wong, Brenda L., Irina Rybalsky, Karen C. Shellenbarger, Cuixia Tian, Mary A. McMahon, Meilan M. Rutter, Hemant Sawnani, and John L. Jefferies. 2016. "Long-Term Outcome of Interdisciplinary Management of Patients with Duchenne Muscular Dystrophy Receiving Daily Glucocorticoid Treatment." *The Journal of Pediatrics*, December.
- Wu, Joyce Y, Karl C K Kuban, Elizabeth Allred, Frederic Shapiro, and Basil T Darras. 2005. "Association of Duchenne Muscular Dystrophy with Autism Spectrum Disorder." *Journal of Child Neurology* 20 (10): 790–95.

X

- Xu, Su, Da Shi, Stephen J P Pratt, Wenjun Zhu, Andrew Marshall, and Richard M. Lovering. 2015. "Abnormalities in Brain Structure and Biochemistry Associated with Mdx Mice Measured by in Vivo MRI and High Resolution Localized 1H MRS." *Neuromuscular Disorders* 25 (10): 764–72.

Y

- Yoshioka, M, T Okuno, Y Honda, and Y Nakano. 1980. "Central Nervous System Involvement in Progressive Muscular Dystrophy." *Archives of Disease in Childhood* 55 (8): 589–94.
- Young, H K, B A Barton, S Waisbren, Dale L Portales, M M Ryan, R I Webster, and K N North. 2008. "Cognitive and Psychological Profile of Males with Becker Muscular Dystrophy." *J.Child Neurol.* 23 (2): 155–62.

Z

- Zamani, Gholamreza, Morteza Heidari, Reza Azizi Malamiri, Mahmoud Reza Ashrafi, Mahmoud Mohammadi, Reza Shervin Badv, Seyed Ahmad Hosseini, et al. 2016. "The Quality of Life in Boys with Duchenne Muscular Dystrophy." *Neuromuscular Disorders* 26 (7): 423–27.

- Zeisel, Steven H, and Kerry-Ann da Costa. 2009. "Choline: An Essential Nutrient for Public Health." *Nutrition Reviews* 67 (11): 615–23.
- Zhang, Y, M Brady, and S Smith. 2001. "Segmentation of Brain MR Images through a Hidden Markov Random Field Model and the Expectation-Maximization Algorithm." *IEEE Transactions on Medical Imaging* 20 (1): 45–57.
- Zhou, X, B Maricque, M Xie, D Li, V Sundaram, E A Martin, B C Koebbe, et al. 2011. "The Human Epigenome Browser at Washington University." *Nat.Methods* 8 (12): 989–90.

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Curriculum Vitae

Nathalie Doorenweerd was born on July 24th 1986 in Vlissingen, The Netherlands. She obtained her bachelor's degree in Bio-Pharmaceutical science from Leiden University, Faculty of Science in 2009. She continued her studies towards a master's in Bio-Pharmaceutical Science specialized in Neuroscience and Pharmaceutical science. Led by an interest in developing new therapeutic strategies targeting the central nervous system and diseases with a cerebral component, she assessed the effect of unilateral epilepsy inducing kainic acid on Blood-Brain-Barrier efflux pump expression and functionality in the rat brain for her first graduation project, which took place at the Department of Pharmacology of Leiden University, Faculty of science under supervision of dr. S. Syvänen, dr. R.A. Voskuyl and dr. E.C. de Lange. This was her first experience with designing and setting up experiments, and included applying for medical ethical committee approval, independently performing surgery and performing all the experiments. For her second graduation project she moved to the Radiology department of Leiden University Medical Centre where she discovered the potential of state-of-the-art imaging techniques in exploring the brain non-invasively *in vivo*. Supervised by dr. L. van der Weerd and D.S. Poole, she helped optimize continuous infusion of manganese to improve image contrast and reduce manganese side-effects in manganese-enhanced magnetic resonance imaging studies in mice. This method was subsequently published in *NeuroImage*.

After obtaining her master's degree in 2011, she started a PhD at Leiden University Medical Center at the C.J. Gorter Center for High Field MRI and her research project 'Brain Imaging and cognition in Duchenne muscular dystrophy' culminated in this thesis. She was given the opportunity to collaborate with experts in the field of very different disciplines and share study results at national and international conferences which was acknowledged with three awards.

Since November 2015 she has been continuing her work on the brain involvement in Duchenne muscular dystrophy by performing longitudinal MRI in an international collaboration between Leiden University Medical Centre, The Netherlands and the John Walton Muscular Dystrophy Research Centre at Newcastle University, United Kingdom.

List of publications

Doorenweerd N, Straathof CS, Dumas EM, Spitali P, Ginjaar IB, Wokke BH, Schrans DG, van den Bergen JC, van Zwet EW, Webb A, van Buchem MA, Verschuuren JJ, Hendriksen JG, Niks EH, Kan HE. *Reduced cerebral gray matter and altered white matter in boys with Duchenne muscular dystrophy*. Ann Neurol. 2014 Sep;76(3):403-11.

Straathof CS, **Doorenweerd N**, Wokke BH, Dumas EM, van den Bergen JC, van Buchem MA, Hendriksen JG, Verschuuren JJ, Kan HE. *Temporalis muscle hypertrophy and reduced skull eccentricity in Duchenne muscular dystrophy*. J Child Neurol. 2014 Oct;29(10):1344-8.

Poole DS, **Doorenweerd N**, Plomp JJ, Mahfouz A, Reinders MJ, van der Weerd L. *Continuous infusion of manganese improves contrast and reduces side effects in manganese-enhanced magnetic resonance imaging studies*. Neuroimage. 2016 Oct 21. pii: S1053-8119(16)30493-1.

Doorenweerd N, Dumas EM, Ghariq E, Schmid S, Straathof CSM, Roest AAW, Wokke BH, van Zwet EW, Webb AG, Hendriksen JGM, van Buchem MA, Verschuuren JGM, Asllani I, Niks EH, van Osch MJP, Kan HE. *Decreased cerebral perfusion in Duchenne muscular dystrophy patients*. Neuromuscul. Disord. 2017 Jan;27(1):29-37

Doorenweerd N, Hooijmans MT, Schubert SA, Webb AG, Straathof CSM, van Zwet EW, van Buchem MA, Verschuuren JJGM, Hendriksen JGM, Niks EH, Kan HE. *Proton magnetic resonance spectroscopy indicates preserved cerebral biochemical composition in Duchenne muscular dystrophy patients*. J. Neuromuscul. Dis. 2017;4(1):53-58

Doorenweerd N¹, Mahfouz A¹, van Putten M, Kaliyaperumal R, 't Hoen PAC, Hendriksen JGM, Aartsma-Rus AM, Verschuuren JJGM, Niks EH, Reinders MJT, Kan HE, Lelieveldt BPF. *Timing and localization of human dystrophin isoform expression provide insights into the cognitive phenotype of Duchenne muscular dystrophy*. Under review

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