

Tay-Sachs Gene Therapy Consortium Publications

1. Porter, B.F., Lewis, B.C., Edwards, J.F., Alroy, J., Zeng, B.J., Torres, P.A., Bretzlaff, K.N. and Kolodny, E.H. (2011) Pathology of GM2 Gangliosidosis in Jacob Sheep. *Vet Pathol* 48: 807 originally published online 1 December 2010
2. Torres, P.A., Zeng, B.J., Porter, B.F., Alroy, J., Horad, F., Horak, J., Kolodny, E.H. (2010) Tay-Sachs Disease in Jacob Sheep. *Molecular Genetics and Metabolism* 101 357-363
3. Roos, J.C., Lachmann, R.H., Carpenter, R.H., Cox, T.M. (2011) Latency vs. Saccadic Parameters in Lysosomal Trials. *Ophthalmology* 118(4): 794.
4. Sargeant, T.J., Wang, S., Bradley, J., Smith, N.J., Raha, A.A., McNair, R., Ziegler, R.J., Cheng, S.H., Cox, T.M., Cachón-González, M.B. (2011) Adeno-Associated Virus-Mediated Expression of β -Hexosaminidase Prevents Neuronal Loss in the Sandhoff Mouse Brain. *Hum Mol Genet.* 20(22):4371-80. Epub Aug 18.
5. Smith, N.J., Winstone, A.M., Stellitano, L., Cox, T.M., Verity, C.M. (2012) GM2 Gangliosidosis in a UK Study of Children with Progressive Neurodegeneration: 73 Cases Reviewed. *Dev Med Child Neurol.* 54(2):176-82. Epub 2011 Nov 24
6. Cachón-González, M.B., Wang, S.Z., McNair, R., Bradley, J., Lunn, D., Ziegler, R., Cheng, S.H., Cox, T.M. (2012) Gene Transfer Corrects Acute GM2 Gangliosidosis--Potential Therapeutic Contribution of Perivascular Enzyme Flow. *Mol Ther.* 20(8):1489-500. Epub Mar 27
7. Bley, A.E., Giannikopoulos, O.A., Hayden, D., Kubilus, K., Tifft, C.J., Eichler, F.S. (2011) Natural History of Infantile G(M2) Gangliosidosis. *Pediatrics* 128(5):e1233-41
8. Sargeant, T.J., Drage, D.J., Wang, S., Apostolakis, A.A., Cox, T.M., Cachón-González, M.B. (2012) Characterization of Inducible Models of Tay-Sachs and Related Disease. *PLoS Genet.* Sep;8(9):e1002943. Epub 2012 Sep 20.
9. Cox, T.M., Cachón-González, M.B. (2012) The Cellular Pathology of Lysosomal Diseases. *J Pathol* 226(2):241-54.
10. Bradbury, A.M., Brunson, B.L., McCurdy, V.J., Gray-Edwards, H., Seyfried, T.N., Cox, N.R., Baker, H.J., Sena-Esteves, M., Martin, D.R. (2013) Dramatic Therapeutic Response of Intracranial AAV Gene Therapy in Feline GM2 Gangliosidosis. *Gordon Research Seminar and Conference: Lysosomal Diseases*, Lucca (Barga), Italy: April 13-19, 2013
11. Bradbury, A.M., McCurdy, V.J., Johnson, A.K., Gray-Edwards, H., Brunson, B.L, Randle, A.N, Cox, N.R., Sena-Esteves, M., Martin, D.R. Intracranial AAV Gene Therapy Extends the Life Span of GM2 Gangliosidosis Cats >Four-Fold with No Clinical Evidence of Vector Toxicity. (2013) *Sixteenth Annual Meeting of the American Society of Gene and Cell Therapy*, Salt Lake City, UT: May 15-18, 2013.

12. McCurdy, V.J., Gray-Edwards, H., Randle, A.N, Bradbury, A.M., Johnson, A.K., Beadlescomb, P.M., Morrison, N.E., Hwang, M., Sena-Esteves, M., Martin, D.R. (2013) Dramatic Phenotypic Improvement after Adeno-Associated Virus Gene Therapy in a Feline Model of Sandhoff Disease. *Sixteenth Annual Meeting of the American Society of Gene and Cell Therapy*, Salt Lake City, UT: May 15-18, 2013.
13. Gray-Edwards, H. L., Hubbard, S. M., Randle, Hwang, M., Bradbury, A. M., McCurdy, V. J., Wilson, D. U., Hudson, J. A., Whitlock, B.K., Sartin, J. L., Salibi, N., Beyers, R. J., Denney, T. S., Johnson, J. A., Riddell, K. P., Cox, N. R., Sena-Estevez, M., Martin, D. R. (2013) Amelioration of Neurologic Disease after AAV-Gene Therapy in a Sheep Model of Tay-Sachs Disease. *Sixteenth Annual Meeting of the American Society of Gene and Cell Therapy*, Salt Lake City, UT: May 15-18, 2013.
14. McCurdy, V.J., Randle, A.N, Gray-Edwards, H., Bradbury, A.M., Johnson, A.K., Beadlescomb, P.M., Sena-Esteves M., Martin D.R. (2013) Significant Therapeutic Benefit after Post-Symptomatic Gene Therapy in a Feline Model of GM2 Gangliosidosis. *Sixteenth Annual Meeting of the American Society of Gene and Cell Therapy*, Salt Lake City, UT: May 15-18, 2013.
15. Gray-Edwards, H., Bradbury, A.M., McCurdy, V.J., Wilson, D. Brunson, B., Holland, M., Salibi, N., Beyers, R, Denny, T., Randle, A.N., Johnson, A.K., Cox, N.R., Sena-Esteves, M., Martin, D.R. (2013) Marked Attenuation of Neurologic Disease and Emergence of Peripheral Disease After Brain-Directed Gene Therapy in a Feline Model of Sandhoff Disease. (2013) *Gordon Research Seminar and Conference: Lysosomal Diseases* April 13-19, 2013
16. Bradbury, A.M., Cochran, J.N., McCurdy, V.J., Johnson, A.K., Brunson, B.L., Gray-Edwards, H., Leroy, S.G., Hwang, M., Randle, A.N., Jackson, L.S., Morrison, N.E., Baek, R.C., Seyfried, T.N., Cheng, S.H., Cox, N.R., Baker, H.J., Cachón-González, M.B., Cox, T.M., Sena-Esteves, M., Martin, D.R. (2013) Therapeutic Response in Feline Sandhoff Disease Despite Immunity to Intracranial Gene Therapy. (2013) *Molecular Therapy* Vol. 21 No. 7 July 2013
17. Weismann, C.M., Ferreira, J., Maitland, S., Su, Q., Guangping, G., Sena-Esteves, M. (2013) Intravenous Administration of AAV9-mβgal in a GM1 Gangliosidosis Mouse Model Expresses Enzyme in the CNS and Delays Disease Onset with Gender Differences. *ASGCT Meeting*, May 15-18, 2013
18. Golebiowski, D., van der Bom, I.M.J., Su-Kown, C., Maitland, S., Kühn, A.L., Bishop, N., Curren, E., Silva, N. Miller, A., Westmoreland, S., Gounis, M., Martin, D.R., Asaad, W., Sena-Esteves, M. (2013) Safety Study of Intracranial AAVrh8-Mediated Gene Transfer of beta-Hexosaminidase in Non-Human Primates. *ASGCT Meeting*, May 15-18, 2013
19. Erger, K.E., Mandel, R.J., Castleman, W.L., Campbell-Thompson, M., Abernathy, C., Getz, M.P., Cossette, T.L., Schneider, L., Brown, R., Sena-Esteves, M., Conlon, T.J. (2013) Biodistribution and Toxicology Studies of AAV2/rh8-CBA-β-Hexosaminidase after Intracranial Delivery in C57/Bl6 Mice. *ASGCT Meeting*, May 15-18, 2013