Rett Syndrome, MECP2 Duplication, and Rett-related Disorders Consortium

Book Chapters

1. Percy A. Rett syndrome: clinical and molecular update. *Current Opinion in Pediatrics.* Vol 162004:670-677.
2. Percy A. Rett syndrome. In: Maria B, ed. *Current Management in Child Neurology*. 3rd ed. Hamilton, Ontario: BC Decker; 2005.
3. Percy A. Rett syndrome: clinical update and future prospects. In: Rubin IL, Crocker AC, eds. *Medical Care for Children and Adults with Developmental Disabilities*. 2nd ed. Baltimore: Paul H. Brookes Publishing 2006:171-178.
4. Percy A. Rett syndrome. In: McMillan J, Feigin RD, DeAngelis C, Jones MD, eds. *Oski's Pediatrics: Principles & Practice*. 5th ed. Philadelphia: Lippincott Williams & Wilkins; 2006:2361-2363.
5. Tarquinio D, Percy A. Rett disorder. In: Hollander E, Loklevzon A, Coyle JT, eds. *APPI Textbook of Autism Spectrum Disorder*2008.
6. Neul J. Rett syndrome and MECP2-related disorders. *Autism Spectrum Disorders*: Oxford University Press; 2009.
7. Percy A, Lane J. Rett syndrome. In: Maria B, ed. *Current Management in Child Neurology.* Vol 375-382. Hamilton, Ontario: BC Decker; 2009.
8. Calfa G, Pozzo-Miller L, Percy A. Rett Syndrome: On Clinical and Genetic Features and Experimental Models Based on MECP2 Dysfunction. In: Powell C, Monteggia L, eds. The Autisms: Molecules to Model Systems. NY: Oxford University Press; 2013:57-90.
9. Chapleau C, Lane J, Pozz-Miller L, Percy A. Rett Syndrome: A Model of Genetic Neurodevelopmental Disorders. In: Puiu M, ed. *Genetic Disorders*: InTech; 2013.

Abstracts Presented at Conferences

1. Van den Veyver I, Amir R, Fang P, Yu Z, Glaze D, Percy A, Zoghbi H, Roa B. Mutations in the newly discovered coding exon 1 of MECP2 are a rare cause of classic Rett syndrome. Paper presented at: ASHG 54th Annual Meeting; October, 2004; Toronto.
2. Neul J, Glaze D, Percy A, Lane J, Barrish J. Specific *MECP2* mutations confer different severity in Rett syndrome. Paper presented at: Child Neurology Society 34th Annual Meeting2005; Los Angeles, CA.
3. Fang P, Ward P, Glaze D, Van den Veyver I, Percy A, Zoghbi H, Roa B. Comprehensive clinical testing of the MECP2 gene for Rett syndrome. Paper presented at: American College of Medical Genetics Annual Meeting; March, 2005.
4. Roa B, Ward P, Glaze D, Neul J, Van den Veyver I, Percy A, Zoghbi H, Fang P. Clinical molecular testing for rett syndrome: comprehensive analysis for point mutations and large rearrangements in the MECP2 gene. Paper presented at: Rett Syndrome Research Foundation Annual Meeting; Jun 27-29, 2005; Chicago, IL.

5. Fang P, Ward P, Berry S, Irons M, Chong B, Van den Veyver I, Neul J, Percy A, Glaze D, Zoghbi H, Roa B. MECP2 gene rearrangements in female and male patients with features of Rett syndrome. Paper presented at: American Society of Human Genetics 55th Annual Meeting; October, 2005; Salt Lake City, UT.
6. Percy A. Summary of clinical trials experience in Rett syndrome. Paper presented at: Clinical Trials In Rett Syndrome International Workshop; May 29-31, 2006; San Francisco, CA.
7. Percy A. Gene therapy in Rett syndrome. Paper presented at: Clinical Trials In Rett Syndrome International Workshop; May 29-31, 2006; San Francisco, CA.
8. Percy A. Rett syndrome: current status and new directions. Paper presented at: 10th Annual International Child Neurology Congress; June, 2006; Montreal, Canada.
9. Percy A. Syndromic forms of mental retardation, neurogenetics for the practitioner. Paper presented at: R. O. Brady Lecture in Neurogenetics; Nov 10, 2006; New York, NY.
10. Percy A. Rett syndrome and MECP2: understanding the genotype-phenotype correlations. Paper presented at: National Society of Genetic Counselors; Nov 13, 2006; Nashville, TN.
11. Tarquinio D, Lane J, Percya A. The natural history of Rett syndrome: phenotypic differences in classical rett syndrome are associated with specific MECP2 mutations. Paper presented at: Rare Disease Clinical Research Network Conference on Clinical Research for Rare Diseases; September, 2007; Bethesda, MD.
12. Tarquinio D, Motil K, Glaze D, Skinner S, Neul J, Annese F, Barrish J, Geerts S, Lane J, Percy A. Growth charts for Rett syndrome: birth to 18 years of age. Paper presented at: Child Neurology Society meeting2008.
13. Peters SU, Tavyev J, Zhang F, Zoghbi H. An emerging behavioral phenotype in female carriers with MECP2 duplications: implications for the broad autism phenotype. Paper presented at: The Keystone Symposia: Toward a Pathophysiology of Autism; February 24, 2008; Santa Fe, NM.
14. Kirby R, Percy A, Lane J, Glaze D, Skinner S, MacLeod P, Barrish J, Annese F. Longevity in Rett syndrome: probing the North American database. Paper presented at: Child Neurology Society meeting; November, 2008.
15. Tarquinio D, Motil K, Glaze D, Skinner S, Neul J, Annese F, Barrish J, Geerts S, Lane J, Percy A. Growth charts for Rett syndrome: birth to 18 years of age. Paper presented at: American Academy of Neurology annual meeting; April, 2009; Seattle, WA.
16. Glaze D, Percy A, Skinner S, Motil K, Neul J, Barrish J, Lane J, Geerts S, Annese F, Graham J, McNair L. Natural history of Rett syndrome: epilepsy. Paper presented at: Pediatric Academic Society meeting; May 2-9, 2009.
17. Percy A, Lee H-S, Glaze D, Skinner S, Motil K, Neul J, Barrish J, Lane J, Geerts S, Annese F, Graham J, McNair L. Profiling scoliosis in Rett syndrome. Paper presented at: Pediatric Academic Society meeting; May 2-9, 2009.

Journal Articles

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2. Percy AK, Lane JB. Rett syndrome: model of neurodevelopmental disorders. *J. Child Neurol.* Sep 2005;20(9):718-721. PMID: 16225824

3. Tofil NM, Buckmaster MA, Winkler MK, Callans BH, Islam MP, Percy AK. Deep sedation with propofol in patients with Rett syndrome. *J. Child Neurol.* Mar 2006;21(3):210-213. PMID: 16901422

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