Rett Syndrome Program Publications
2015-2017

GGC faculty names in bold

- Tarquinio DC, Hou W, Neul JL, Lane JB, Barnes KV, O'Leary HM, Bruck NM, **Kaufmann WE**, Motil KJ, Glaze DG, **Skinner SA, Annese F, Baggett L,** Barrish JO, Geerts SP, Percy AK (2015) Age of diagnosis in Rett syndrome: patterns of recognition among diagnosticians and risk factors for late diagnosis. Pediatr Neurol 52: 585-591.
- Herrera JA, Ward CS, Pitcher MR, Percy AK, Skinner S, **Kaufmann WE**, Glaze DG, Wehrens XHT, Neul JL (2015) Treatment of cardiac arrhythmias in Rett syndrome with sodium channel blocking antiepileptic drugs. Dis Model Mech 8: 363-371.
- O'Driscoll OM, Lima MP, **Kaufmann WE**, Bressler JP (2015) Methyl CpG binding protein 2 deficiency enhances expression of inflammatory cytokines by sustaining NF-κB signaling in myeloid derived cells. J Neuroimmunol 283: 23-29.
- Olson HE, Tambunan D, LaCoursiere C, Goldenberg M, Pinsky R, Martin E, Ho E, Khwaja O, **Kaufmann WE***, Poduri A***** (2015) Mutations in epilepsy and intellectual disability genes in patients with features of Rett syndrome. Am J Med Genet 167A: 2017-2025.
- Tarquinio DC, Hou W, Neul JL, **Kaufmann WE**, Glaze DG, Motil KJ, **Skinner SA,** Lee H-S, Percy AK (2015) The changing face of survival in Rett syndrome. Pediatr Neurol 53: 402-411.
- O'Leary HM, Marschik PB, Khwaja OS, Ho E, Barnes KV, Clarkson TW, Bruck NM, **Kaufmann WE** (2017) Detecting autonomic response to pain in Rett syndrome. Dev Neurorehabil 20: 108-114.
- Barnes KV, Coughlin FR, O'Leary HM, Bruck N, Bazin GA, Beinecke EB, Walco AC, Cantwell NG, **Kaufmann WE** (2015) Anxiety-like behavior in Rett syndrome: characteristics and assessment by anxiety scales. J Neurodevelop Disord 7: 30.
- LeBlanc JJ, DeGregorio G, Centofante E, Vogel-Farley VK, Barnes K, **Kaufmann WE**, Fagiolini M, Nelson CA (2015) Visual evoked potentials detect cortical processing deficits in Rett syndrome. Ann Neurol 78: 775-786.
- Killian JT, Lane JB, Lee H-S, Pelham JH, **Skinner SA, Kaufmann WE**, Glaze DG, Neul JL, Percy AK (2016) Caretaker quality of life in Rett syndrome: disorder features and psychological predictors. Pediatr Neurol 58: 67-74.
- Sajan SA, Jhangiani SN, Muzny DM, Gibbs RA, Lupski JR, Glaze DG, **Kaufmann WE**, **Skinner SA, Annese F, Friez MJ,** Lane J, Percy AK, Neul JL. (2017) Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. Genet Med 19: 13-19.
- Jefferson A, Leonard H, Siafarikas A, Woodhead H, Fyfe S, Ward LM, Munns C, Motil K, Tarquinio D, Shapiro JR, Brismar T, Ben-Zeev B, Bisgaard AM, Coppola G, Ellaway C, Freilinger M, Geerts S, Humphreys P, Jones M, Lane J, Larsson G, Lotan M, Percy A, Pineda M, **Skinner S,** Syhler B, Thompson S, Weiss B, Witt Engerström I, Downs J (2016) Clinical Guidelines for Management of Bone Health in Rett Syndrome Based on Expert Consensus and Available Evidence. PLoS One: 11(2):e0146824.
- Tarquinio DC, Hou W, Berg AT, **Kaufmann WE**, Lane J, **Skinner SA,** Motil KJ, Neul JL, Percy AK, Glaze DG (2017) Longitudinal course of epilepsy in Rett syndrome and related disorders. Brain 140: 306-318.
- Ward CS, Huang T-W, Herrera JA, Samaco RC, Pitcher MR, Herron A, Skinner SA, **Kaufmann WE**, Glaze DG, Percy AK, Neul JL (2016) Loss of MeCP2 causes urological dysfunction and contributes to death by kidney failure in mouse models of Rett syndrome. PLoS One 11: e0165550.
- Lane JB, Salter AR, Jones NE, Cutter G, Horrigan J, **Skinner SA, Kaufmann WE**, Glaze DG, Neul JL, Percy AK (2017) Assessment of Caregiver Inventory for Rett Syndrome. J Autism Dev Disord 47: 1102-1112.
- Killian JT, Lane JB, Lee H, **Skinner SA, Kaufmann WE**, Glaze DG, Neul JL, Percy AK (2017) Scoliosis in Rett Syndrome: progression, comorbidities, and predictors. Pediatr Neurol: Feb 07.
- **Kaufmann WE**, Stallworth JL, Everman DB, Skinner SA (2016) Neurobiologically-based treatments in Rett syndrome: opportunities and challenges. Expert Opin Orphan Drugs 4: 1043-1055.
- Marschik PB, Pokorny FB, Peharz R, Zhang D, O'Muircheartaigh J, Roeyers H, Bölte S, Spittle AJ, Urlesberger B, Schuller B, PhD; Poustka L, Ozonoff S, Pernkopf F, Pock T, Tammimies K, Enzinger C, Krieber M, Tomantschger I, Bartl-Pokorny KD, Sigafoos J, Roche L, Esposito G, Gugatschka M, Nielsen-Saines K, Einspieler C, Walter E **Kaufmann WE** & the BEE-PRI study group (2017) A novel way to measure and predict development: A heuristic approach to facilitate the early detection of neurodevelopmental disorders. Curr Neurol Neurosci Rep 17: 43.

- Clarkson T, LeBlanc J, DeGregorio G, Vogel-Farley V, Barnes K, **Kaufmann WE**, Nelson CA (2017) Adapting the Mullen Scales of Early Learning for a Standardized Measure of Development in Children with Rett Syndrome. Intellect Dev Disabil: in press.