

Publications by Elwyn Staff



Fragile X Syndrome

- Greco, C.M., Navarro, C.S., Hunsaker, M.R., Maezawa, I., Shuler, J.F., Tassone, F., Delany M., Au, J.W., Berman, R.F., Jin, L., Schumann C., Hagerman, P.J., & Hagerman, R.J. (2011). Neuropathologic features in the hippocampus and cerebellum of three older men with fragile X syndrome. *Molecular Autism*, 2(2) [Doi:10.1186/2040-2392-2-2]
- Bourgeois, J., Coffey, S., Rivera, S.M., Hessler, D., Gane, L.W., Tassone, F., Greco, C., Finucane, B., Nelson, L., Berry-Kravis, E., Grigsby, J., Hagerman, P.J., and Hagerman, R.J. (2009). Fragile X premutation disorders – Expanding the psychiatric perspective. *Journal of Clinical Psychiatry*, 70(6), 852-62.
- McConkie-Rosell, A., Abrams, L., Finucane, B., Cronister, A., Gane, L.W., Coffey, S.M., Sherman, S., Nelson, L.M., Berry-Kravis, E., Hessler, D., Chiu, S., Street, N., Vatave, A., and Hagerman, R.J. (2007). Recommendations from multi-disciplinary focus groups on cascade testing and genetic counseling for fragile X-associated disorders. *Journal of Genetic Counseling*, 16,593-606.
- Berry-Kravis, E., Abrams, L., Coffey, S.M., Hall, D.A., Greco, C., Gane, L.W., Grigsby, J., Bourgeois, J.A., Finucane, B., Jacquemont, S., Brunberg, J.A., Zhang, L., Lin, J., Tassone, F., Hagerman, P.J., Hagerman, R.J., and Leehey, M.A. (2007). Fragile X-associated tremor / ataxia syndrome: Clinical features, genetics, and testing guidelines. *Movement Disorders*. 22,2018-30.
- McConkie-Rosell, A., Finucane, B., Cronister, A., Abrams, L., Bennett, R.L., and Pettersen, B.G. (2005). Genetic counseling for fragile X syndrome: Updated recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling*. 14,249-269.
- Finucane, B. (2005): What's in a name? Symptoms versus causes in the diagnostic age. *Exceptional Parent*, 35,26-28.
- Finucane, B., McConkie-Rosell, A., and Cronister, A. (2002): *Fragile X Syndrome: A Handbook for Families and Professionals*. San Francisco: National Fragile X Foundation.
- Finucane, B. and Simon, E.W. Diagnostic alphabet soup: Demystifying genetic and behavioral labels. *The Foundation Quarterly*. Winter 2001, National Fragile X Foundation.
- Finucane, B. and Cronister, A. (2000). Genetic counseling. In J.D. Weber (Ed.), *Children with Fragile X Syndrome*. Woodbine, NJ: Woodbine Publishers.
- Dykens, E., Ort, S., Cohen, I., Finucane, B., Spiridigliozzi, G., Lachiewicz, A., Reiss, A., Freund, L., Hagerman, R., and O'Connor, R. (1996). Trajectories and profiles of adaptive behavior in males with fragile X syndrome: Multicenter studies. *Journal of Autism and Developmental Disabilities*, 26,287-301.
- Finucane, B. (1996). Should all pregnant women be offered carrier testing for fragile X syndrome? *Clinical Obstetrics and Gynecology*, 39,772-782.
- Simon, E.W. and Finucane, B. (1996). Facial emotion identification in males with fragile X syndrome. *Neuropsychiatric Genetics*, 67,77-80.
- Simon, E.W., Rappaport, D.A., Papka, M., and Woodruff-Pak, D.S. (1995). Fragile X and Down syndrome: Are there syndrome-specific cognitive profiles at low IQ levels?, *Journal of Intellectual Disability Research*, 39, 326-330.
- Ramos, F.J., Eunpu, D.L., Finucane, B., Pfenner, E.G. (1993). Direct DNA testing for fragile X syndrome. *American Journal of Diseases in Childhood*, 147,1231-1235.
- Finucane, B., Jaeger, E., Dunn, E., and Scott, C.I. (1992). A study of color vision in fragile X syndrome. *American Journal of Medical Genetics*, 42,184-186.
- Finucane, B., Scott, C., and Kurtz, M. (1991). Concurrence of dominant piebald trait and fragile X syndrome (letter). *American Journal of Human Genetics*, 48,815.
- Dykens, E., Hodapp, R., Ort, S., Finucane, B., Shapiro, L., and Leckman, J. (1989). The trajectory of cognitive development in males with fragile X syndrome. *Journal of the American Academy of Child and Adolescent Psychiatry*, 28,422-426.

Smith-Magenis Syndrome

- Finucane, B. and Haas-Givler, B. (2009): Smith-Magenis syndrome: Genetic basis and clinical Implications. *Journal of Mental Health Research in Intellectual Disabilities*, 2,134-148.
- Elsea, S.H. and Finucane, B. (March 2009). Smith–Magenis syndrome. In: *Encyclopedia of Life Sciences (ELS)*. Chichester: John Wiley & Sons, Ltd Chichester <http://www.els.net/> [DOI: 10.1002/9780470015902.a0021428]

- Finucane, B. (2008). Embracing the inner toddler in people with Smith-Magenis syndrome. *Spectrum*, Summer 2008, PRISMS.
- Haas-Givler, B. (2008). Prevention versus intervention: Effective behavioral strategies for children with Smith-Magenis syndrome. *Spectrum*, Spring 2008, PRISMS.
- Edelman, E.A., Girirajan, S., Finucane, B., Patel, P., Lupski, J.R., Smith, A.C.M., and Elsea, S.H. (2007). Gender, genotype, and phenotype differences in Smith-Magenis syndrome: A meta-analysis of 105 cases. *Clinical Genetics*, 71,1-11.
- Smith, A.C.M., Allanson, J., Elsea, S.H., Finucane, B., Haas-Givler, B., Gropman, A., Johnson, K., Lupski, J.R., Magenis, R.E., Potocki, L., and Solomon, B. (2006). Smith-Magenis syndrome, In *GeneReviews*, Seattle, WA: University of Washington, GeneTests: Medical Genetics Information Resource (online database).
- Simon, E. W. (2004). Smith Magenis syndrome. In D. Griffiths, R. King, and R. Ryan (Eds.), *Demystifying Syndromes: Clinical and Educational Implications of Common Syndromes Associated with Persons with Intellectual Disabilities*, Kingston, NY: NADD Press.
- Finucane, B. (2003). The long road to diagnosis, by way of a needle in a haystack. *Spectrum*, Summer / Fall 2003, PRISMS.
- Slager, R.E., Newton, T.L., Vlangos, C.N., Finucane, B., and Elsea, S.H. (2003). Mutations in retinoic acid induced 1 (RAI1) associated with Smith-Magenis syndrome phenotype in patients without a detectable 17p11.2 deletion. *Nature Genetics*, 33,466-468.
- Finucane, B., Dirrgl, K., and Simon, E.W. (2001). Characterization of self-injurious behaviors in children and adults with Smith-Magenis syndrome. *American Journal on Mental Retardation*, 106,52-58.
- Finucane, B. (2001). Psychiatric diagnoses as they relate to Smith-Magenis syndrome. *Spectrum*, Winter 2001, PRISMS.
- Finucane, B. and Simon, E.W. (1999). Genetics and dual diagnosis: Smith-Magenis syndrome. *NADD Bulletin*, 1,8-10.
- Dykens, E., Finucane, B., and Gayley, C. (1997). Brief report: Cognitive and behavioral profiles in persons with Smith-Magenis syndrome. *Journal of Autism and Developmental Disabilities*, 27,203-211.
- Finucane, B. (1997). Smith-Magenis syndrome (letter). *Ophthalmology*, 104,732.
- Elsea, S.H., Purandare, S.M., Adell, R.A., Juyal, R.C., Davis, J.G., Finucane, B., Magenis, R.E., and Patel, P.I. (1997). Definition of the critical interval for Smith-Magenis syndrome. *Cytogenetics and Cell Genetics*, 79,276-281.
- Trask, B.J., Mefford, H., van den Engh, G., Massa, H.F., Juyal, R.C., Finucane, B., Abuelo, D.N., Witt, D.R., Magenis, E., Baldini, A., Greenberg, F., Lupski, J.R., and Patel, P.I. (1996). Quantification by flow cytometry of chromosome 17 deletions in Smith-Magenis syndrome. *Human Genetics*, 98,710-718.
- Juyal, R.C., Finucane, B., Shaffer, L.G., Lupski, J.R., Greenberg, F., and Scott, C.I. (1995). Apparent mosaicism for del(17)(p11.2) ruled out by FISH in a Smith-Magenis syndrome patient. *American Journal of Medical Genetics*, 59,406-407.
- Haas-Givler, B. and Finucane, B.M. (1995). What's a teacher to do? Classroom strategies that enhance learning for children with Smith-Magenis syndrome. *Spectrum*, Winter / Spring 1995, PRISMS.
- Haas-Givler, B. (1994). Observations on the behavioral and personality characteristics of children with Smith-Magenis syndrome. *Spectrum*, Summer 1994, PRISMS.
- Finucane, B., Konar, D., Haas-Givler, B., Kurtz, M., and Scott, C.I. (1994). The spasmodic upper body squeeze: A characteristic behavior in Smith-Magenis syndrome. *Developmental Medicine and Child Neurology*, 36,78-83.
- Finucane, B., Kurtz, M., Babu, V., and Scott, C.I. (1993). Mosaicism for deletion 17p11.2 in a boy with the Smith-Magenis syndrome. *American Journal of Medical Genetics*, 45,47-449.
- Finucane, B., Jaeger, E., Kurtz, M., Weinstein, M., and Scott, C.I. (1993). Eye abnormalities in the Smith-Magenis contiguous gene deletion syndrome. *American Journal of Medical Genetics*, 45,443-446.

Down Syndrome

- Berry, G.T., Wang, Z.J., Dreha, S.F., Finucane, B.M., and Zimmerman, R.A. (1999). In vivo brain myo-inositol levels in children with Down syndrome. *Journal of Pediatrics*, 135,94-97.
- Pietrini, P., Dani, A., Furey, M. L., Alexander, G. E., Freo, U., Grady, C. L., Mentis, M. J., Mangot, D., Simon, E. W., Horwitz, B., Haxby, J. V., and Schapiro, M. B. (1997). Low glucose metabolism during brain stimulation in older Down's syndrome subjects at risk for Alzheimer's disease prior to dementia. *American Journal of Psychiatry*, 154,1063-1069.

- Simon, E. W., Agriesti, M., and Rappaport, D. A. (1995). Memory performance in adults with Down syndrome. *Australia and New Zealand Journal of Developmental Disabilities*, 20,113-125.
- Woodruff-Pak, D.S., Papka, M., and Simon, E.W. (1994). Eyeblink classical conditioning in Down's syndrome, fragile X syndrome, and normal adults over and under age 35. *Neuropsychology*, 8,14-24.
- Papka, M., Simon, E.W., and Woodruff-Pak, D. (1994). Longitudinal investigation of eyeblink conditioning in adults with Down syndrome. *Aging and Cognition*, 1,89-104.

Other Genetic Conditions

- Simon, E.W., Haas-Givler, B., and Finucane, B. (2010): A Longitudinal Follow-up Study of Autistic Symptoms in Children and Adults with Duplications of 15q11-13. *Am J Med Genet Part B* 153B:463-467.
- Feuk, L., Kalervo, A., Lipsanen-Nyman, M., Skaug, J., Nakabayashi, K., Finucane, B., Hartung, D., Innes, M., Kerem, B., Nowaczyk, M.J., Rivlin, J., Roberts, W., Senman, L., Summers, A., Szatmari, P., Wong, V., Vincent, J.B., Zeesman, S., Osborne, L.R., Cardy, J.O., Kere, J., Scherer, S.W., Hannula-Jouppi, K. (2006). Absence of a paternally inherited FOXP2 gene in developmental verbal dyspraxia. *American Journal of Human Genetics*, 79,965–972.
- Finucane, B. (2004). Williams syndrome. In D. Griffiths, R. King, and R. Ryan (Eds.), *Demystifying Syndromes: Clinical and Educational Implications of Common Syndromes Associated with Persons with Intellectual Disabilities*, Kingston, NY: NADD Press.
- Haas-Givler, B. (2004). Education, treatment and Intervention models: Implications for teaching children with idic(15) and related disorders. *The Mirror*, Summer 2004, IDEAS.
- Finucane, B. The growing importance of genetic support groups. *22q and You*. Winter 2002, Children's Hospital of Philadelphia.
- McDonald-McGinn, D.M., Tonnesen, M.K., Laufer-Cahana, A., Finucane, B., Driscoll, D.A., Emanuel, B.S., and Zackai, E.H. (2001). Phenotype of the 22q11.2 deletion in individuals identified through an affected relative: Cast a wide FISHing net! *Genetics in Medicine*, 3,23-29.
- Finucane, B. and Simon, E.W. Educational and behavioral diagnoses in children with a 22q deletion. *22q and You*, Winter / Spring 2001, Children's Hospital of Philadelphia.
- Simon, E. W., Finucane, B., and Rineer, S. (2000). Autistic symptoms in isodicentric 15 syndrome: Response to Wolpert et al. *American Journal of Medical Genetics (Neuropsychiatric Genetics)*, 96,427-428.
- Rineer, S., Finucane, B., Simon, E.W. (1998). Autistic symptoms among children and young adults with isodicentric chromosome 15. *American Journal of Medical Genetics (Neuropsychiatric Genetics)*, 81,428-433.
- Kurtz, M.B., Finucane, B., Hyland, K., Bottiglieri, T., Sherwood, W.G., and Bennett, M.J. (1994). Detection of metabolic disorders among selectively screened people with idiopathic mental retardation. *Mental Retardation*, 32,328-333.
- Finucane, B., Kurtz, M., and Scott, C.I. (1992). New mental retardation syndrome with deafness, distinct facial appearance, and skeletal anomalies. *American Journal of Medical Genetics*, 43,844-847.

Genetics and Developmental Disabilities

- Dent, K.M., Harper, C., Kearney, L., Lieber, C., and Finucane, B. 2011. Embracing the unique role of genetic counselors: Response to the commentary by Madeo et al., *American Journal of Medical Genetics Part A*.
- Finucane, B. (2010). Genetic Counseling for Women with Intellectual Disabilities. In B. LeRoy, P.M. Veach, and D.M. Bartels (Eds.), *Genetic Counseling Practice: Advanced Concepts and Skills*. Hoboken, N.J.: John Wiley and Sons, Inc.
- Levitas, A., Dykens, E., Finucane, B., and Kates, W.R. (2007). Behavioral phenotypes of genetic disorders. In R. Fletcher, E. Loschen, C. Stavrakaki, and M. First (Eds.), *Diagnostic Manual – Intellectual Disability: A Textbook of Diagnosis of Mental Disorders in Persons with Intellectual Disability*. Kingston, N.Y.: NADD Press.
- Finucane, B., Haas-Givler, B., and Simon, E.W. (2003). Genetics, mental retardation, and the forging of new alliances. *American Journal of Medical Genetics (Part C, Seminar in Medical Genetics)*, 117C,66-72.
- Finucane, B. (2002). Prenatal testing and disability rights (book review). *American Journal on Mental Retardation*, 107,490-491.
- Dykens, E., Hodapp, R., and Finucane, B. (2000). *Genetics and Mental Retardation Syndromes: A New Look at Behavior and Interventions*. Baltimore: Brookes Publishing.
- Simon, E.W. and Finucane, B. (2000). Genetics and developmental disabilities: Autistic disorder. *NADD Bulletin*, 3,31-33.
- Finucane, B. (1998). *Working with Women who have Mental Retardation: A Genetic Counselor's Guide*. Elwyn, PA: Elwyn, Inc.

- Simon, E.W. and Finucane, B. (1998). Etiology and dual diagnosis: Notes on a biologically-based syndromic approach. *NADD Bulletin*, 1,63-65.
- Finucane, B. (1998). Acculturation in women with mental retardation and its impact on genetic counseling. *Journal of Genetic Counseling*, 7,31-47.
- Finucane, B. (1996). *What's so Special About Genetics? A Guide for Special Educators*. Elwyn, PA: Elwyn, Inc.
- Finucane, B. (1991). Long-term counseling at a center for individuals with mental retardation. *Perspectives in Genetic Counseling*, 13,2.

Miscellaneous

- Lichten, W. L. and Simon, E. W. (in press). Response to Bellini's Commentary on Lichten and Simon. *Intellectual and Developmental Disabilities*.
- Lichten, W. L. and Simon, E. W. (2007). Defining Mental Retardation: A Matter of Life or Death. *Intellectual and Developmental Disabilities*, 45,335-346.
- Simon, E. W., Whitehair, P. M., and Toll, D.M. (1996). A case study: Follow-up assessment of facilitated communication. *Journal of Autism and Developmental Disabilities*, 26,9-18.
- Simon, E. W., Blubaugh, K. M., and Pippidis, M. (1996). Substituting traditional antipsychotics with Risperidone for individuals with mental retardation. *Mental Retardation*, 34,359-366.
- Simon, E. W. & Rappaport, D. A. (1996). Specific response blocking, exposure and differential reinforcement eliminate disrobing: A case study. *British Journal of Developmental Disabilities*, 82,70-74.
- Simon, E. W., Rosen, M. & Ponpipom, A. (1996). Age and IQ as predictors of emotion identification in adults with mental retardation. *Research in Developmental Disabilities*, 17,383-389.
- Simon, E. W., Toll, D.M., & Whitehair, P. M. (1995). Keeping facilitated communication in perspective. *Mental Retardation*, 33,338-339.
- Simon, E. W., Rosen, M., Grossman, E., and Pratoski, E. (1995). The relationships among facial emotion recognition, social skills and quality of life. *Research in Developmental Disabilities*, 16,383-391.
- Rosen, M., Simon, E. W., and Mckinsey, L. (1995). A subjective measure of quality of life. *Mental Retardation*, 33,31-34.
- Simon, E. W., Whitehair, P., and Toll, D.M. (1994). A naturalistic investigation into facilitated communication. *Journal of Autism and Developmental Disabilities*, 24,647-657.