Brenda M Finucane

BUSINESS ADDRESS Geisinger Health System
Autism & Developmental Medicine Institute
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CURRENT POSITION

January 2013 to present: Associate Director and Professor,
Autism & Developmental Medicine Institute
Professor, Genomic Medicine Institute
Geisinger Health System
Danville, Pennsylvania

- Responsible for administrative duties and leadership initiatives related to strategic planning, direction, fiscal management, operations, staff recruitment, supervision, and program marketing for ADMI's integrated clinical and research program
- Senior investigator on original and collaborative research projects involving genetic etiologies of developmental brain disorders
- Licensed genetic counselor, clinical provider of diagnostic genetic assessments and genetic counseling

PRIOR EMPLOYMENT

September 1985 to December 2012: Executive Director, Genetic Services
Genetic Counselor and Clinical Investigator
Elwyn, Inc.
Elwyn, Pennsylvania

- Established and directed the department of Genetic Services at a large, nonprofit human services organization serving adults and children with developmental disabilities
- Developed a business model for the department based on fee-for-service contracts; grant-funded initiatives; multicenter subcontracts; and targeted development campaigns
- Served in an executive leadership capacity for the organization and its Division of Research and Health Services
- Senior clinical investigator on original and collaborative research projects related to genetic counseling; behavioral and cognitive phenotyping of genetic syndromes; and targeted pharmaceutical trials for fragile X syndrome and other genetic disorders

July 1980 - August 1983: Peace Corps volunteer
Zaire (Democratic Republic of Congo), Africa

- Taught Science and English as a second language to students in a remote equatorial village; established a regional health outreach and education program for pregnant women and infants
- Served as a consultant to the government of Zaire on health curriculum development for the national school system

EDUCATION

May 1985: MS in Human Genetics
Sarah Lawrence College
Bronxville, New York

May 1980: BA in Biology / English Literature (Double Major)
New York University
New York, New York

LICENSURE / SPECIALTY BOARDS

1987 Genetic Counseling, American Board of Medical Genetics
1993 Genetic Counseling (charter certification), American Board of Genetic Counseling
2006 Recertification, Genetic Counselor, American Board of Genetic Counseling
2011 Recertification, Genetic Counselor, American Board of Genetic Counseling
2014 Licensure, Genetic Counseling, Pennsylvania State Board of Medicine

LEADERSHIP / SERVICE

- *American Board of Genetic Counseling*: Board of Directors, 2002 - 2006; Chair, Credentials Committee, 2002-2004; Item Writer, 2002 national certification exam
- *Duplication 15q Alliance* (Cofounder): Scientific Advisory Committee, 1994 – present
- *International Fragile X Clinical and Research Consortium:* Membership and Infrastructure Committee, 2009 - present
- *International 22q11.2 Deletion Syndrome Foundation*: Board of Directors, 2006 – 2011; Professional Advisory Committee, 2006 - present
- *Mid-Atlantic Regional Human Genetics Network*: Chair, Education Committee, 1996-99; Chair, Special Projects Subcommittee, 1995-96.
- *National Fragile X Foundation*: Board of Directors, 2014 - present; International Scientific and Clinical and Advisory Committee, 1997 – present.
- *National Fragile X Advocate*: Professional Advisory Board, 1995-97
- *National Society of Genetic Counselors*: Immediate Past President, 2013; President, 2012; President-elect, 2011; Chair, Nominating Committee, 2013; Genetic Counseling Advanced Degree Task Force, 2012; Board of Directors, 2008 – 2009.
- *NIH National Institute of Child Health and Human Development*: program project grant reviewer (Fragile X syndrome), 2014.
- *Parents and Researchers Interested in Smith-Magenis Syndrome* (Cofounder): Professional Advisory Board, 1993 – present.
- *Pennsylvania Genetic Counselors State Licensure Committee* (Cofounder); Officer, 2006 – 2012
- *Positive Exposure:* Board of Directors, 2015

EDITORSHIPS AND EDITORIAL BOARDS

- *Journal of Genetic Counseling*: Editorial Board: 1998 – 2003; 2008 – 2012
- *Journal of Genetic Counseling:* Guest Editor, Special Issue on Developmental Disabilities, December 2012

MANUSCRIPT REVIEWER

American Journal on Intellectual and Developmental Disabilities
American Journal of Medical Genetics
Intellectual and Developmental Disabilities
Journal of Autism and Developmental Disorders
Journal of Genetic Counseling
Journal of Intellectual Disability Research
Journal of Mental Health Research in Intellectual Disabilities

SELECTED HONORS / AWARDS

- Duplication 15q Alliance: *Service and Leadership Award* (2006)
- National Fragile X Foundation: *Dorn Education and Awareness Award* (2002)
- Mid-Atlantic Regional Human Genetics Network: *Gold Chromosome Award* (Best Grant Project 1995)

GRANT SUPPORT

Active

R01MH107431-01 (Co-I: Finucane B – 10% effort; PI: Martin CL) 9/15/2015-6/30/2020
NIH, *Dimensional Analysis of Developmental Brain Disorders Using an Online, Genome-first Approach*
Current year direct costs: $495,507

Bucknell Geisinger Research Initiative (MPI: Finucane B – 10% effort; Mitchel A) 1/1/15-12/31/17
Multisensory Integration in Klinefelter and Turner Syndromes Direct costs: $87,319

RO1 MH074090 (Co-I: Finucane B – 40%; MPI: Martin CL, Ledbetter DH) 5/1/16 - 4/30/21
NIH/ NIMH, *Gene Dosage Imbalance in Neurodevelopmental Disorders* Direct Costs: $489,959

Previous Funding

FRAXA Research Foundation (Postdoctoral fellow: King M – 90% effort; PI: Finucane B) 5/1/14 - 4/30/16
Analysis of Developmental Brain Dysfunction (DBD) in Families with Fragile X Syndrome: A New Approach to Clinical Trial Triage Direct costs: $45,000

NEU_CH_7911: FDA Clinical Trial (Co-I: Finucane B – 10% effort; PI – Challman TD) 1/13/16 – 6/30/16
A Randomized, Placebo-controlled Study to Investigate the Efficacy and Safety of Circadin® to Alleviate Sleep Disturbances in Children with Neurodevelopmental Disabilities Direct costs (estimated): $122,660

NEU-2566-FXS-001: FDA Clinical Trial (Co-I: Finucane B–10% effort; PI–Challman TD) 1/13/15 – 12/31/15
A Randomized, Double-Blind, Placebo-Controlled, Parallel-Group, Fixed-Dose Study of NNZ-2566 in Fragile X Syndrome Direct costs (estimated): $81,884

RO1 MH074090 (Co-I: Finucane B - 20% effort; PI: Ledbetter DH) 07/01/14 – 04/30/16
NIH/NIMH, *Gene Dosage Imbalance in Neurodevelopmental Disorders* Current year direct costs: $527,154

Simons Foundation (Co-I: Finucane B – 10% effort; PI: Martin CL) 6/01/13 – 7/31/15
Identifying the Gene in 17q12 Responsible for Neuropsychiatric Phenotypes Direct costs: $145,833

Dup15q Alliance (Research Assistant: Lusk L – 10% effort; PI: Finucane B) 7/1/14 - 6/30/15
Analysis of Family-reported Data from the Dup15q Registry Direct costs: $3,649

Bucknell Geisinger Research Initiative (MPI: Finucane B – 10% effort; Mitchel A) 6/1/13 - 5/31/14
Genetic Variation in CNTNAP2 and Correlation with Multisensory Integration in the Broader Autism Phenotype Direct costs: $50,000

PRISMS, Inc. (PI: Finucane B – 33% effort) 1/1/14 - 3/31/14
On the Road to Success with SMS: A Smith-Magenis Guidebook for Schools Direct costs: $20,000

Martha W Rogers Trust (PI: Finucane B – 20% effort) 1/1/08 – 12/31/08
Developmental Asynchrony in Individuals with Smith-Magenis Syndrome Direct costs: $29,000

Martha W Rogers Trust (PI: Finucane B – 20% effort) 1/1/06 – 12/31/07
Knowledge and Attitudes among Special Educators about Genetic Causes of Developmental Disabilities
Direct costs: $62,000

Mid-Atlantic Regional Human Genetics Network (PI: Finucane B - 10% effort) 1/1/95 – 12/31/95
What's So Special About Genetics? A Guide for Special Educators Direct costs: $10,000

National Society of Genetic Counselors' Jane Engelberg Memorial Fellowship Award 1/1/94 - 12/31-94
Genetic Counseling and Reproductive Decision-making in Women with Mental Retardation
Direct costs: $25,000

PEER REVIEWED PUBLICATIONS

Ajay, O.J.,, Smith, E.J., Viangteeravat, T., Huang, E.Y., Nagisetty, N.S., Urraca, N., Lusk, L., **Finucane, B**., Arkilo, D., Young, J., Jeste, J., Thibert, R., The Dup15q Alliance, & Reiter, L.T. (2017). Multisite semiautomated clinical data repository for duplication 15q syndrome: study protocol and early uses*. JMIR Research* Protocols 6(10):e194. doi:10.2196/resprot.7989

Brendal, M.A., King, K.A., Zalewski, C.K., **Finucane, B.M.**, Introne, W., Brewer, C.C., & Smith, A.C.M. (2017). Auditory phenotype of Smith–Magenis Syndrome. *Journal of Speech, Language, and Hearing Research.* Epub ahead of print, doi.org/10.1044/2016_JSLHR-H-16-0024

Finucane, B., Lincoln, S., Bailey, L., & Martin, C.L. (2016). Prognostic dilemmas and genetic counseling for

prenatally-detected fragile X expansions. *Prenatal Diagnosis.* 36,1-6. doi:10.1002/pd.4963

Mitchel, M.W., Moreno-De-Luca, D., Myers, S.M., **Finucane, B**., Ledbetter, D.H., & Martin, C.L. 2016 Dec 8. 17q Recurrent Delection Syndrome. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): Univ of Washington, Seattle; 1993-2016. Available from: https://www.ncbi.nlm.nih.gov/books/NBK401562/

Finucane, B. & Myers, S.M. (2016). Genetic counseling for autism spectrum disorder in an evolving theoretical landscape. *Current Genetic Medicine Reports*. Epub ahead of print, doi:10.1007/s40142-016-0099-9

Liu, J.A., Hagerman, R.J., Miller, R.M., Craft, L.T., **Finucane, B.**, Tartaglia, N., Berry-Kravis, E.M., Sherman, S.L., Kidd, S.A., & Cohen, J. (2016). Clinicians' experiences with the fragile X clinical and research consortium. *American Journal of Medical Genetics Part A*, 9999A:1–6.

Finucane, B.M., Lusk, L., Arkilo, D., et al. 15q Duplication Syndrome and Related Disorders. 2016 Jun 16. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016. Available from: http://www.ncbi.nlm.nih.gov/books/NBK367946/

Finucane, B., Challman, T.D., Martin, C.L., & Ledbetter, D.H. (2016). Shift happens: Family background influences clinical variability in genetic neurodevelopmental disorders. *Genetics in Medicine*, 18(4),302-304. doi:10.1038/gim.2015.92

Nagy, R., Peay, H., Hicks, M., Kloos, J., Westman, R., Conway, L., **Finucane, B.**, Fitzpatrick, J., Gordon, E., Ramos, E., Sekhon-Warren, J., Silver, J., Walton, C., & Reiser, C. (2014). Genetic counselors' and genetic counseling students' attitudes around the clinical doctorate and other advanced educational options for genetic counselors: A report from the Genetic Counseling Advanced Degree Task Force. Journal of Genetic Counseling. doi 10.1007/s10897-014-9785-5

Yrigollen, C.M., Martorell, L., Durbin-Johnson, B., Naudo, M., Genoves, J., Murgia, A., Polli, R., Zhou, L., Barbouth, D., Rupchock, A., **Finucane, B**., Latham, G., Hadd, A., Berry-Kravis, E., & Tassone, F. (2014). AGG interruptions and maternal age affect *FMR1* CGG repeat allele stability during transmission. Journal of Neurodevelopmental Disorders, 6(24),1-12. doi:10.1186/1866-1955-6-24

Conant, K. D., **Finucane, B**., Cleary, N., Martin, A., Muss, C., Delany, M., Murphy, E. K., Rabe, O., Luchsinger, K., Spence, S. J., Schanen, C., Devinsky, O., Cook, E. H., LaSalle, J., Reiter, L. T. and Thibert, R. L. (2014), A survey of seizures and current treatments in 15q duplication syndrome. Epilepsia. doi: 10.1111/epi.12530

Finucane, B., Haas-Givler, B., and Simon, E.W. (2013), Knowledge and perceptions about fragile X syndrome: Implications for diagnosis, intervention, and research. Intellectual and Developmental Disabilities, 51(4), 226-236. doi: 10.1352/1934-9556-51.4.226

Finucane, B., Abrams, L., Cronister, A., Archibald, A.D., Bennett, R.L., & McConkie-Rosell, A.: Genetic counseling and testing for *FMR1* gene mutations: Practice Guidelines of the National Society of Genetic Counselors. Journal of Genetic Counseling, 21(6):752-760, 2012.

Finucane, B.: 2012 National Society of Genetic Counselors presidential address: Maintaining our professional identity in an ever-expanding genetics universe. Journal of Genetic Counseling, 21(1):3-6, 2012, Epub 2011 Dec 23, PMID: 22194035.

Smith, A.C.M., Boyd, K.E., Elsea, S.H**., Finucane, B.M**., Haas-Givler, B., Gropman, A., Laje, G., Magenis, E., & Potocki, L.: Smith-Magenis Syndrome (June 2012) in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle, 1997-2010.

Abrams, L., Cronister, A., Brown, W.T., Tassone, F., Sherman, S.L., **Finucane, B.,** McConkie-Rosell, A., Hagerman, R., Kaufmann, W.E., Picker, J., Coffey, S., Skinner, D., Johnson, V., Miller, R., & Berry-Kravis, E.: Newborn, carrier, and early childhood screening recommendations for fragile X. Pediatrics, 130(6):1126-1135, 2012, Epub 2012 Nov 5, PMID:2312907.

Dent, K.M., Harper, C., Kearney, L., Lieber, C., & **Finucane, B.:** Embracing the unique role of genetic counselors: Response to the commentary by Madeo et al., American Journal of Medical Genetics, Part A 155A:1791-3, 2011. Epub 2011 Jul 7, PMID: 21739.

Simon, E.W., Haas-Givler, B., & **Finucane, B.**: A longitudinal follow-up study of autistic symptoms in children and adults with duplications of 15q11-13. American Journal of Medical Genetics, Part B 153B:463-467, 2010.

Finucane, B. & Haas-Givler, B.: Smith-Magenis syndrome: Genetic basis and clinical implications. Journal of Mental Health Research in Intellectual Disabilities, 2:134-148, 2009.

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., & Leehey, M.A.: Fragile X-associated tremor/ataxia syndrome: Clinical features, genetics, and testing guidelines. Movement Disorders 22:2018-30, 2007.

Edelman, E.A., Girirajan, S., **Finucane, B.,** Patel, P., Lupski, J.R., Smith, A.C.M., & Elsea, S.H.: Gender, genotype, and phenotype differences in Smith–Magenis syndrome: A meta-analysis of 105 cases. Clinical Genetics 71:540-550, 2007.

McConkie-Rosell, A., Abrams, L., **Finucane, B.,** Cronister, A., Gane, L.W., Coffey, S.M., Sherman, S., Nelson, L.M., Berry-Kravis, E., Hessl, D., Chiu, S., Street, N., Vatave, A., & Hagerman, R.J.: Recommendations from multi-disciplinary focus groups on cascade testing and genetic counseling for fragile X-associated disorders. Journal of Genetic Counseling 16:593-606, 2007.

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.: Absence of a paternally inherited *FOXP2 g*ene in developmental verbal dyspraxia. American Journal of Human Genetics 79:965–972, 2006.

McConkie-Rosell, A., **Finucane, B.,** Cronister, A., Abrams, L., Bennett, R.L., & Pettersen, B.G.: Genetic counseling for fragile X syndrome: Updated recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling 14:249-269, 2005.

Finucane, B., Haas-Givler, B, & Simon, E.W.: Genetics, mental retardation, and the forging of new alliances. American Journal of Medical Genetics, Part C (Seminar in Medical Genetics), 117C:66-7, 2003.

Slager, R.E., Newton, T.L., Vlangos, C.N., **Finucane, B.,** & Elsea, S.H.: 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, 2003.

Finucane, B., Dirrgl, K., & Simon, E.W.: Characterization of self-injurious behaviors in children and adults with Smith-Magenis syndrome. American Journal on Mental Retardation, 106:52-58, 2001.

McDonald-McGinn, D.M., Tonnesen, M.K., Laufer-Cahana, A., **Finucane, B.,** Driscoll, D.A., Emanuel, B.S., & Zackai, E.H.: Phenotype of the 22q11.2 deletion in individuals identified through an affected relative: Cast a wide FISHing net! Genetics in Medicine 3:23-29, 2001.

Berry, G.T., Wang, Z.J., Dreha, S.F., **Finucane, B.M.,** & Zimmerman, R.A.: In vivo brain myo-inositol levels in children with Down syndrome. Journal of Pediatrics 135:94-97, 1999.

Finucane, B.: Acculturation in women with mental retardation and its impact on genetic counseling. Journal of Genetic Counseling, 7:31-47, 1998.

Rineer, S., **Finucane, B.,** & Simon, E.W.: Autistic symptoms among children and young adults with isodicentric chromosome 15. American Journal of Medical Genetics (Neuropsychiatric Genetics) 81:428-433, 1998.

Finucane, B.: Smith-Magenis syndrome (letter). Ophthalmology, 104:732, 1997.

Dykens, E., **Finucane, B.,** & Gayley, C.: Brief report: Cognitive and behavioral profiles in persons with Smith-Magenis syndrome. Journal of Autism and Developmental Disorders 27:203-211, 1997.

Elsea, S.H., Purandare, S.M., Adell, R.A., Juyal, R.C., Davis, J.G., **Finucane, B.,** Magenis, R.E., & Patel, P.I.: Definition of the critical interval for Smith-Magenis syndrome. Cytogenetic Cell Genetics 79:276-28, 1997.

Finucane, B.: Should all pregnant women be offered carrier testing for fragile X syndrome? Clinical Obstetrics and Gynecology, 39:772-782, 1996.

Dykens, E., Ort, S., Cohen, I., **Finucane, B.,** Spiridigliozzi, G., Lachiewicz, A., Reiss, A., Freund, L., Hagerman, R., & O'Connor, R.: Trajectories and profiles of adaptive behavior in males with fragile X syndrome: Multicenter studies. Journal of Autism and Developmental Disorders 26:287-301, 1996.

Simon, E.W. & **Finucane, B.**: Facial emotion identification in males with fragile X syndrome, American Journal of Medical Genetics (Neuropsychiatric Genetics) 67:77-80, 1996.

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., & Patel, P.I.: Quantification by flow cytometry of chromosome-17 deletions in Smith-Magenis syndrome. Human Genetics 98:710-718, 1996.

Elsea, S.H., Juyal, R.C., Jiralerspong, S., **Finucane, B.M.,** Pandolfo, M., Greenberg, F., Baldini, A., Stover, P., & Patel, P.I.: Haploinsufficiency of cytosolic serine hydroxymethyltransferase in the Smith-Magenis syndrome. American Journal of Human Genetics 57:1342-1350, 1995.

Juyal, R.C., **Finucane, B.,** Shaffer, L.G., Lupski, J.R., Greenberg, F., & Scott, C.I.: 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, 1995.

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Kurtz, M.B., **Finucane, B.,** Hyland, K., Bottiglieri, T., Sherwood, W.G., & Bennett, M.J.: Detection of metabolic disorders among selectively screened people with idiopathic mental retardation. Mental Retardation 32:328-333, 1994.

Finucane, B., Jaeger, E., Kurtz, M., Weinstein, M., & Scott, C.I.: Eye abnormalities in the Smith-Magenis contiguous gene deletion syndrome. American Journal of Medical Genetics 45:443-446, 1993.

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Finucane, B., Kurtz, M., & Scott, C.I. New mental retardation syndrome with deafness, distinct facial appearance, and skeletal anomalies. American Journal of Medical Genetics 43:844-847, 1992

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BOOKS AND CHAPTERS

Levitas, A., **Finucane, B**., Simon, E.W., Schuster, M., Kates, W.R., Olszewski, A., Dykens, E.M. & Danker, N. Behavioral phenotypes of neurodevelopmental disorders. In R. Fletcher, J. Barnhill, & S.A. Cooper (Eds.) Diagnostic Manual – Intellectual Disability: A Textbook of Diagnosis of Mental Disorders in Persons with Intellectual Disability, 2nd Edition. Kingston, NY: NADD Press, 2016.

Finucane, B.: Williams syndrome. In D.M. Griffiths & S.L. Watson (Eds.) Demystifying Syndromes II: Clinical and Educational Implications of Common Syndromes Associated with Persons with Intellectual Disabilities. Kingston, NY: NADD Press, 2016

Braden, M. & **Finucane, B.** Fragile X syndrome. In D.M. Griffiths & S.L. Watson (Eds.) Demystifying Syndromes II: Clinical and Educational Implications of Common Syndromes Associated with Persons with Intellectual Disabilities. Kingston, NY: NADD Press, 2016.

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Simon, E.W., Haas-Givler, B., & **Finucane, B**. An introduction to genetic intellectual disability syndromes: Basic concepts and applications for applied behavior analysis professionals. In Griffiths, D., Condillac, R.A., & Legree, M. (Eds.) Genetic Syndromes and Applied Behavior Analysis. London: Jessica Kingsley Publishers, 2014.

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OTHER PUBLICATIONS

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