CURRICULUM VITAE

NAME: Luis Fernando Escobar, M.D., M.S.

PRESENT ADDRESS:

Medical Director - Staff Neonatologist
Medical Genetics and Neurodevelopmental Center
St. Vincent Hospitals and Health Services

EDUCATION:

San Carlos University of Guatemala, C.A. Biology, **B.S.**, 1975–1978
San Carlos University of Guatemala, C.A. Medicine, **M.D.**, 1978–1984
Indiana University School of Medicine, Indianapolis, Indiana, Genetics, **M.S.**, 1985–1988

RESIDENCY:

Indiana University School of Medicine, Indianapolis, Indiana, Pediatric Internship and Residency Training, 7/01/90 – 6/30/93

FELLOWSHIPS:

Indiana University School of Medicine, Indianapolis, Indiana, Department of Oral-Facial Genetics and Medical Genetics, Postdoctoral Fellow, 8/01/85 6/30/88
Regional Genetics Center, Memorial Hospital of South Bend, Indiana, Postdoctoral Fellow, 8/01/89 – 6/30/90
Indiana University School of Medicine, Indianapolis, Indiana, Department of Pediatrics, Section of Neonatal-Perinatal Medicine, Postdoctoral Fellow, 7/01/93 –6/30/98.

ACADEMIC APPOINTMENTS:

Assistant Research Scientist, Indiana University School Of Medicine, Department of Oral Facial Genetics 1987-1989
Visiting Scholar, University of Notre Dame, Department Of Biological Sciences 1989-1990.
Lecturer in Pediatrics, Indiana University School of Medicine, Department of Pediatrics 1993-1994.
Visiting Scientist, University of Auckland School of Medicine, Animal Research. New Zealand, 1997.
Adjunct Assistant Professor of Medical & Molecular Genetics, Indiana University Department of Medical and Molecular Genetics, Indianapolis, Indiana 1999 - 2013

TEACHING EXPERIENCE:

Indiana University:
93KN700 senior elective in Neonatology-lecturer and

Supervising fellow for clinical instruction 1993-1998.

K710 Pediatric clerkship-lecturer and supervising fellow 1993-1998.

Neonatal Resuscitation skills practicum for Pediatric House staff, 1993-1998

St. Vincent Hospital:

Clinical Lecturer and Supervising staff for Family Practice, OB/GYN and Pediatric house staff, 1998-Present

CLINICAL TRIALS EXPERIENCE:

Principal Investigator: 10 Week double blind placebo controlled study to evaluate the efficacy and safety of Donepezil Hydrochloride in the treatment of the cognitive dysfunction exhibited by children with Down syndrome. Pfizer, New York. Protocols: 1059, 1060 and 1061 (2010)

Principal Investigator: A double blind placebo controlled, randomized withdrawal study of the safety and efficacy of Memantine in Pediatric Patients with autism, asperger's disorder or pervasive developmental disorder. Forest Laboratories, Protocols: MEM-MD-68, 69, 91

LICENSURE:

State of Indiana

AWARDS AND HONORS:

Physician of the Year Award, March of Dimes, Indianapolis 2015

Virginia Wagner Memorial Award. American Academy of Pediatrics, Indiana Chapter, legislative efforts, volunteerism and lifetime service to children, 2014

Indiana Minority Business & Professional Achievers Award, Indiana Center for Leadership Development, 2012

Edwin L Gresham Award- Advancing care of Infants. American Academy of Pediatrics, Indiana Chapter. 2009

March of Dimes-Charlene Lugard Award, 1999

Minority Supplemental Award, NIH 1998

National Research Service Award (NIDR) 1 F32 DE05456-10A1, 1986

Oral Facial Genetics Training Grant DE 7043 (Postdoctoral Fellowship), 1985

American Field Service Recognition, Honduras, Central America, Work toward world peace, 1978

Guatemala Ministry of Health Fellowship for the senior year of Medical Education 1984

SOCIETIES:

American Society of Human Genetics, 1985 – (Active)
American Academy of Pediatrics, 1990 - (Active)
American College of Medical Genetics 2000 – (Active)
Society of Craniofacial Genetics, 1986 - 2005
Vice-President 1995 – 1997
President 1997 – 1998
American Institute of Ultrasound in Medicine, 1985 - 1989
Sigma Xi, the Scientific Research Society, 1988 - 1999

COMMUNITY WORK:

1. Chairman State Advisory Committee for Genetic Services, Indiana Department of Health. 2014-present
2. AAP EHDI Liaison, District V. 2013-present
3. Member of the State Advisory Committee for Genetic Services, Indiana Department of Health. Sub-committee chair. 2000 – 2013
4. Member Advisory Committee of Indiana Early Hearing Detection and Intervention (EHDI Program, Indiana State Department of Health 2005 – Present
5. AAP Pediatric Champion. Early Hearing Detection and Intervention Program, Indiana Department of Health 2010-present
6. Member of Board of Directors for the Indiana Muscular Dystrophy Family Foundation 2006 - 2009
7. Member of Board of Directors Indiana Perinatal Network 2007- 2008
8. Trustee-Board of Directors Indiana Down Syndrome Foundation, 2001-2005
9. Member of the Central Indiana First Steps Local Planning & Coordinating Council, 2007- 2008

NATIONAL INVOLVEMENT:

1. Consultant, Molecular and Clinical Genetics Panel of the Medical Devices Advisory Committee, Center for Devices and Radiological Health, Food and Drug Administration. Department of Health and Human Services.

2. Member for the Molecular and Clinical Genetics Advisory Committee FDA/CDRH/ODE, Washington, USA

MAJOR SCIENTIFIC INTEREST:

Hypoxic–Ischemic Cell Injury
Craniofacial Genetics
Neonatal-Perinatal Medicine
Clinical Genetics and Dysmorphology

PROFESSIONAL ACTIVITIES:

Editorial Review Group Chair for Medical Genetics, Doody's Book Review Service
Clinical Council Member, National Marfan Syndrome Foundation

HOSPITAL APPOINTMENTS:

Staff Neonatologist
St. Francis Hospital of Indianapolis 1996-1998
Staff Neonatologist
Women's Hospital of Indianapolis 1999-present
Staff Neonatologist and Director of Medical Genetics
St. Vincent Hospital and Health Care system, 1998-present
Staff Pediatrician & Medical Geneticist
Clarian North Medical Center, 2005 – 2009

ADDITIONAL EXPERTISE:

VitalStim Therapy Provider – Pediatric Focus, Provider 81185
Charlotte NC, 4/2009.
Modified Barium Swallow, CIAO Charlotte NC, June 30, 2009.
CPR & AED Healthcare Provider, Am Heart Association, 2/16/2018
ACLS Provider, Am Heart Association 12/12/2018

BOOK CHAPTERS:

1. **Escobar LF**: Malformations of the facial bones. In Human Malformations. Stevenson R, Hall J, and Goodman RM, Editors. Oxford University Press, Vol II, pp 629-649, 1993.

2. **Escobar LF,** Weaver DD: Auriculo-osteodysplasia. Birth Defects Encyclopedia. Blackwell Scientific publications pp 212-213, 1990.

3. **Escobar LF**, Weaver DD: Ocular colobomas. Birth Defects Encyclopedia. Blackwell Scientific Publications, pp 950-951, 1990.

4. **Escobar LF,** Weaver DD: CHARGE association. Birth Defects Encyclopedia. Blackwell Scientific publications, pp 308-309, 1990.

5. **Escobar LF**, Weaver DD, Bixler D: Urorectal Septum Malformation Sequence. Birth Defects Encyclopedia. Blackwell Scientific Publications, pp 1736-1737, 1990.

6. Gripp K and **Escobar LF**: Facial Bones. In Human Malformations and Related Anomalies. Stevenson R and Hall J. Second Edition. Pp 267-292, 2006

7. Davis AS and **Escobar LF**: Down syndrome. Pediatric Neuropsychology. Davis (Ed.). First Edition. Chapter 63, Springer Publishing Company, 2010

BIBLIOGRAPHY:

1. **Escobar, LF**: Low birth weight-birth intervals relationship in 1000 normal pregnancies at Social Security and Roosevelt Hospitals in Guatemala. Medical Doctor Thesis, San Carlos University of Guatemala School of Medicine. October 1984.

2. Flores MV, **Escobar LF**, Alvarado J, Aguilar JL, Menendez H, Alvarado C, Gonzales E: Malnutrition in Guatemala. A symposium in honor of Lic. Dante Liano. Colegio Salesiano Don Bosco, Guatemala, C.A.

3. Weaver DD, **Escobar LF**: Twenty-four ways to have children. American Journal of Medical Genetics, 26:737-740, 1987.

4. **Escobar LF**, Weaver DD, Bixler D, Hodes ME, Mitchell M: The urorectal septum malformation sequence. American Journal of Disease in Children, 141:1021-1024, 1987.

5. **Escobar LF**, Bixler D, Padilla LM, Weaver DD: Fetal craniofacial morphometrics in utero at 16 weeks of gestation. American Journal of Human Genetics 41:A273, 1987.

6. **Escobar LF**, Bixler D, Weaver DD, Sadove M: Antley-Bixler syndrome from a prognostic perspective: Report of a case and review of the literature. Proceedings of the Greenwood Genetics Center 7:206, 1988.

7. **Escobar LF**, Bixler D, Sadove M, Bull M: Antley-Bixler syndrome from a prognostic perspective: report of a case and review of the literature. American Journal of Medical Genetics 29:829-836, 1988.

8. **Escobar LF**, Weaver DD, Haun R: Ureteric bud-mesenchyme interaction failure? Report of a case and Embryological review. Proceedings of the Greenwood Genetic Center 7:183, 1988.

9. **Escobar LF**, Bixler D, Padilla LM, Weaver DD: Fetal craniofacial morphometrics in utero. Evaluation at 16 weeks of gestation. Obstetrics and Gynecology, 72:677-679, 1988.

10. Reed T, **Escobar LF**: Dermatoglyphics in the Antley-Bixler Syndrome. Journal of Dysmorphology and Clinical Genetics, 2:6-8, 1988.

11. **Escobar LF**, Bixler D, Bull MJ, Bader P, Weaver DD: A provisionally unique-pattern syndrome: craniosynostosis-knee dislocation, short fingers and toes (follow-up). Proceedings of the Greenwood Genetic Center 8:209-210, 1989.

12. **Escobar LF**, Bixler D, and Weaver DD: Mozart ears and amniotic bands disruption sequence in the Roberts syndrome. Proceedings of the Greenwood Genetic Center 8:212, 1989.

13. **Escobar LF**, Weaver DD, Win J: Sirenomelia. Birth Defects Encyclopedia. Blackwell Scientific Publications, pp 1541-1542, 1990.

14. **Escobar LF**, Weaver DD: Caudal regression syndrome. Birth Defects Encyclopedia. Blackwell Scientific Publications, pp 296-297, 1990.

15. **Escobar LF**, Weaver DD, Bixler D, Hodes ME, Mitchell M: The urorectal septum malformation sequence. Year Book of Urology, Year Book of Medical Publishers, Inc. Chicago, pp 233-234, 1988.

16. **Escobar LF**, Weaver DD, Bixler D, Hodes ME, Mitchell M: The urorectal septum Sequence. Proceedings of the Greenwood Genetics Center 6:149, 1988.

17. **Escobar LF**, Weaver DD: Thoracic myelomeningocele without vertebral arch defects. Proceedings of the Greenwood Genetic Center 8:191, 1989.

18. **Escobar LF**, Bixler D, Weaver DD, Bull MJ, Bader P: Larsen syndrome and Craniosynostosis: Idaho syndrome? Journal of Dysmorphology and Clinical Genetics 3:24-27, 1989.

19. **Escobar LF**, Bixler D, Weaver DD, Padilla LM, Golichowski A: Bone Dysplasias: the prenatal diagnostic challenge. American Journal of Medical Genetics 36:488-494, 1989.

20. **Escobar LF**, Bixler D, Padilla LM, Weaver DD: Fetal Craniofacial Morphometrics: In utero evaluation at 16 weeks gestation. Journal of Diagnostic Medical Sonography 1:25, 1989.

21. **Escobar LF**, Bixler D, Padilla LM: Determination of Fetal Craniofacial Dysmorphology in utero by morphometric analysis with ultrasound. American Journal of Physical Anthropology 78:218, 1989.

22. **Escobar LF**, Bixler D, Padilla LM: Fetal cephalometry by ultrasound: New standardized values. American Journal of Human Genetics 45:A258, 1989.

23. Carlin ME, **Escobar LF**, Ward R: A reassessment of Beckwith-Wiedemann syndrome (BWS). American Journal of Human Genetics 47:A50, 1990.

24. Ward R, **Escobar LF**, Carlin ME, Haines JL: Quantitative analysis of the face in the Beckwith-Wiedemann Syndrome and detection of minimally affected gene carriers. American Journal of Human Genetics 47:A82, 1990.

25. **Escobar LF**, Bixler D, Padilla LM, Weaver DD, Williams CJ: A morphometric analysis of the fetal craniofacies by ultrasound: fetal cephalometry. Journal of Craniofacial Genetics and Developmental Biology 10:19-27, 1990.

26. **Escobar LF**, Bixler D: Fetal Cephalometry. Year Book of Speech, Language and Hearing, Eds. Bernthal JE, Hall JW, Tomblin JB. Mosby-Year Book, Inc., Chicago, pp 37-38, 1991.

27. **Escobar LF**, Bixler D, Padilla LM, Weaver DD, Williams, CJ: Morphometric analysis of the fetal craniofacies: fetal cephalometry. The Journal of Craniofacial Surgery 2(3):159.

28. **Escobar LF**, Bixler D, Padilla LM: Quantitation of craniofacial anomalies in utero: Fetal alcohol and Crouzon syndromes and Thanatophoric dysplasia. American Journal of Medical Genetics 45:25-29, 1993.

29. **Escobar LF**, Sokol G, Torres W, Faught PR, Davis M: Neonatal presentation of Kenny-Caffey syndrome with megaloencephaly, thymus-spleen hypoplasia, absent parathyroid and intrauterine growth retardation. (In-Press).

30. Wheeler PG, Weaver DD, Obeime, MO, Vance GH, Bull MJ, **Escobar LF**: Urorectal Septum Malformation Sequence: Report of Thirteen Additional Cases and Review of the Literature. American Journal of Medical Genetics 73:456-462, 1997.

31. **Escobar LF**, Boyle DW, Yoder MC, Liechty EA: Brain polyubiquitin up-regulation in hypoxic ischemic encephalopathy (HIE) and intrauterine growth retardation. Pediatric Research 43(4):318A, 1997.

32. Liechty EA, Boyle DW, **Escobar LF**, Denne SC: Regulation of Phenylalanine (Phe) and Tyrosine (Tyr) kinetics in fetal sheep. Pediatric Research 41(4):236A, 1997.

33. **Escobar LF**, Bixler D, Padilla LM, Liechty EA: Fetal craniofacial pattern variability index: the measurement of fetal craniofacial dysmorphology. Pediatric Research 41(4):147A, 1997.

34. **Escobar LF,** Liechty EA: Late gestational vascular disruptions inducing craniofacial anomalies: a Fetal model. Pediatric Research 41(4):60A, 1997.

35. Liechty EA, Boyle DW, **Escobar LF**, Denne SC: Ovine fetal leucine kinetics during graded infusion of rhIGF-I. Pediatric Research 41(4):69A, 1997.

36. **Escobar LF** and Liechty E: Late Craniofacial Vascular disruptions inducing craniofacial anomalies: A fetal lamb model. Journal of Craniofacial Genetics and Developmental Biology 18:159-163, 1998.

37. **Escobar LF**, Jabs EW, Melnick M: Society of Craniofacial Genetics 1997 Annual Meeting Abstracts. Journal of Craniofacial Genetics and Developmental Biology 18:110, 1998.

38. Moore ES, Ward RE, **Escobar LF** and Carlin ME: Heterogeneity in Wiedeman-Beckwith Syndrome Anthropometric Evidence. American Journal of Medical Genetics. 90:283-290, 2000.

39. **Escobar LF,** Hiett KA, Marnocha A, and Cohen S: Phenotypic variability of the Muenke syndrome

in twins with an identical de-novo C749G mutation in the FFR-3 gene and prenatal exposure to nortriptyline. Proceedings of the Greenwood Genetic Center 23:149, 2004.

40. **Escobar LF**, Heiman M, Steffus L, Harter L: Progressive Postnatal Macrocephaly-Autism a Recognizable syndrome of heterogeneous origin. New evidence: review of 26 patients. Proc of the Greenwood Genet Ctr (24):77, 2005

41. **Escobar LF**, Heiman M, Zimmer D, Careskey H: Urorectal septum malformation sequence: Prenatal Progression, Clinical Report and Embryology Review. Am J Med Gent Part A 143A:2722-2726 (2007).

42. **Escobar LF.** Principles of Molecular Medicine. Shock 28:499 (2007)

43. Ballif B C., Theisen A, Coppinger J, Gowans G, Hersh J., Madan-Khetarpal S, Schmidt K, Tervo R, **Escobar LF**, Friedrich C, McDonald M, Ming J, Zackai E, Carmack C, Bejjani B, Shaffer L. Expanding the Clinical Phenotype of the 3q29 Microdeletion Syndrome and Characterization of The Reciprocal Microduplication. Molecular Cytogenetics 1:8 (2008)

44. **Escobar LF**, Bradburn N, King K: Influence of Prematurity on Developmental Outcomes in Down Syndrome. Proc of the Greenwood Genet Ctr (27):114, (2008)

45. **Escobar LF**: Diagnostic Techniques in Genetics. Medicine & Science in Sports & Exercise. 40(4):777 (2008).

46. **Escobar LF:** Principles and Practice of Medical Genetics. Shock 30(3):339 (2008)

47. **Escobar LF**, Weaver DD. Evaluation of prenatal-onset osteochondrodysplasias by ultrasonography: A persistent challenge 18 years later. Am J Med Genet Part A 149A:1099–1101. (2009).

48. **Escobar LF**, Hiett AK, Marnocha A: Significant Phenotypic Variability of Muenke Syndrome in Identical Twins. Am J Med Genet Part A 149A:1273-1276. (2009).

49. Messiaen L, Yao S, Brems H, Callens T, **Escobar LF**, et al: Clinical and Mutational Spectrum of Neurofibromatosis Type 1–like Syndrome. *JAMA.* 302(19):2111-2118 (2009).

50. **Escobar LF**, El-Khechen D: Autosomal Dominant Chiari I Malformation: Syndrome delineation, Definition of non-neurostructural abnormalities, and Neurobehavioral patterns. Review of 81 Patients. Proc of the Greenwood Genet Ctr, (2009). 28(1):45

51. Moore ES, Ramsey CJ, **Escobar LF**, and Sumners JE: Triplet gestation and developmental outcomes. Am J Obstet Gynecol, (2009). 201(6)

52. Girirajan S, Rosenfeld J, Cooper GM, **Escobar LF**, et al: A recurrent 16p12.1 microdeletion suggests a two-hit model for severe developmental delay. Nature Genetics. 42(3):203-9 (2010)

53. Kishnani PS, Heller JH, Spiridigliozzi GA, Lott I, **Escobar LF**, Richardson S, Zhang R, McRae T: Donepezil for treatment of cognitive dysfunction in children with Down syndrome aged 10-17. Am J Med Genet Part A 152A:3028–3035 (2010).

54. Rosenfeld JA; Lacassie Y; El-Khechen D; **Escobar LF**, et al: New cases and refinement of the critical region in the 1q41q42 microdeletion syndrome. Eur J Med Genet. 2011. 54(1):42-9

55. **Escobar LF**: Medical Genetics (Media review). Medicine & Science in Sports & Exercise, (2011) 43(7):1378

56. Rosenfeld JA, Lacassie Y, El-Khechen D, **Escobar LF,** et al: Exclusion of *DISP1* as a candidate gene for the 1q41q42 microdeletion syndrome. Euro J Med Genet, (2012) 54(1):42-9

57. **Escobar LF**, Wagner S, Wareham J, Tucker M: Neonatal Presentation of Lethal Neuromuscular Glycogen Storage Disease Type IV. Journal of Perinatology. (2012) 32:810-813

58. Girirajan S, Rosenfeld JA, Coe Bradley P, Parikh S, **Escobar LF**: The Phenotypic Heterogeneity of Genomic Disorders and Multiple, Rare CNVs. New England Journal of Medicine (2012). 367(14):1321-31

59. Rosenfeld JA[1], Traylor RN, Schaefer GB, McPherson EW, Ballif BC, Klopocki E, Mundlos S, Shaffer LG, Aylsworth AS, **Escobar LF**; 1q21.1 Study Group. Proximal microdeletions and microduplications of 1q21.1 contribute to variable abnormal phenotypes. Eur J Hum Genet 2012 Jul;20(7):754-61

60. Dabell MP, Rosenfeld JA, Bader P, **Escobar LF**: Investigation of NRXN1 Deletions: Clinical and Molecular Characterization. (2013) Am J Med Genet, Part A999:1-15

61. Hirata H, Nanda I, Van Riesen A, **Escobar LF**: ZC4H2 mutations are associated with arhtrogryposis multiplex congénita and intelectual disability through impairment of central and peripheral synaptic plasticity. Am J Hum Genet, (2013), 92(5):681-95

62. Handrigan G, **Escobar, LF**: Deletions in 16q24.2 are associated with cognitive impairment and congenital renal malformation. J Med Genet (2013), 50(3):163-73

63. Nagamani S, Erez A, **Escobar LF**: Detection of copy-number variation in Aut2 gene targeted exonic Array CGH in patients with developmental delay and autistic spectrum disorders. Euro J of Hum Genet, (2013), 21(3):343-6

64. Girirajan S, Rosenfeld JA, Coe BP, **Escobar LF**, et al: Multiple rare CNVs contribute to the phenotypic heterogeneity of genomic disorders. New England Journal of Medicine (In-press)

65. Tucker M, **Escobar LF:** Cleft Lip/Palate associated with 17p13.3 duplication involving a single candidate gene (YWHAE). Clin Genet, (2014), 85:600-601.

66. Tucker ME, Kalb FM, **Escobar LF:** Infant Spinocerebellar Ataxia Type 27: Early Presentation Due To a 13q33.1 Microdeletion Involving the FGF14 Gene. J Genet Syndr Gene Ther (2013) 4:208. doi: 10.4172/2157-7412.1000208.

67. Foster T, Sumners J, **Escobar LF**: Cognitive Developmental outcomes of triplets. Am J Obstet & Gynecol (2013), 208(1):S212-S213

68. Soster EL, Tucker M, **Escobar LF** and Vance H: Hydranencephaly in a Newborn with a FLVCR2 Mutation and Prenatal Exposure to Cocaine. Birth Defects Research (Part A) and Molecular Teratology (Impact Factor: 2:27). 01/2014:DOI: 10.1002/bdra.

69. Rosenfeld JA, Tucker ME, **Escobar LF** et al: Diagnostic utility of microarray testing of Pregnancy losses. Ultrasound in Ob and Gyn. (2015) 46(4):478-86, DOI:10.1002/uog.14866

70. Trippi JA, **Escobar LF**, Tucker M, Myers E Genetic Diagnosis of Familial Hypercholesterolemia in a Community Lipd Clinic. J of Clin Lipidology (2015), 9(3):427-429

71. Riggs CB, MD[1], **Escobar LF**, Tucker ME: Early-Infantile Galactosialidosis: Clinical and Radiological Findings. J Clin Med Case Reports. (2015);2(2): 4.

72. Rojnueangnit K, Xie J, Gomes A, **Escobar LF**, et al: High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients Carrying NF1 Missense Mutations Affecting p.Arg1809:Genotype-Phenotype Correlation. Hum Mut J (2015), 36(11):1052-1063

74. Yang H, Douglas G, Monaghan KG., Retterer K, Cho MT., **Escobar LF.** *De novo* truncating variants in the *AHDC1* gene encoding the AT-hook DNA-binding motif-containing protein 1 are associated with intellectual disability and developmental delay. Cold Spring Harb Mol Case Studies 1(1): a000562 doi: 10.1101/mcs.a 000562. 2015

75. **Escobar LF**, Tucker M, Bamshad M: A second family with CATSHL syndrome: Confirmatory report of another unique FGFR3 syndrome. Am J Med Genet Am J Med Genet 170:1908-1911, 2016

76. Zak J, Vives V, Szumska D, **Escobar LF,** Tucker M, Aylsworth AS, Dubbs HA, Collins AT, Andrieux J: ASPP2 Deficiency causes features of 1q41q42. Cell Death Differ. 2016 Jul 22. doi: 10.1038/cdd.2016.76.

77, Whitley B, Lam C, Cui H, Haude K, Bai R, **Escobar L**, Hamilton A, Brady L, Tarnopolsky M, Dengle L,

Picker J, Lincoln S, Lackner L, Glass I, Hoppins S: Aberrant Drp1- mediated mitochondrial divisions presents in humans with variable outcomes. Hum Mol Genet, ddy287, https://doi.org/10.1093/hmg/ddy287

RECENT PRESENTATIONS:

1. Hear Indiana. The Genetics of Hearing Loss, October 30, 2009, Indianapolis, Indiana

2. Triplet Gestation and Developmental Outcomes poster presentation at the Society for Maternal Fetal Medicine 30th Annual Meeting, Chicago, IL, February 4, 2010.

3. Ethical Considerations of Lethal Genetic Disorders in Pregnancy. OB/GYN Residency Ethics Day, St. Vincent Women's Hospital, March 10, 2010

4. Negative Effects of Prematurity on Developmental Outcomes in Down Syndrome at age two. L Escobar, W. Pratt, N Bradburn. 2010 ACMG annual Clinical Genetics Meeting, March 24-28, 2010.

5. Further Characterization of the Duplication 16p13.11 Phenotype. Jennifer Kussmann, Escobar LF, Weaver DD et al. 2011 ACMG annual Clinical Genetics Meeting, March 16-20, 2011

6. A case against perfection. OB/GYN Residency Ethics Day, St. Vincent Women's Hospital, March 10, 2010

7. Low Density Lipoprotein Receptor-Related Protein 1B (LRP1B) Gene Anomalies: role in Autism Spectrum Escobar LF, Tucker M, El-khechen D. Annual American College of Medical Genetics Meeting. Vancouver BC, March 16-20, 2011

8. Hearing loss: From Detection to Diagnosis. EHDI National Meeting, March 2012. St. Louis. MO

9. YWHAE gene associated with craniofacial midline development. David W. Smith 33th Annual Workshop on Malformations and Morphogenesis. Lake Laniere, Georgia August, 2012

10. Uncovering The Cause of Autism: From Detection to Diagnosis Clinical Genetic Evaluation And Testing In 393 Patients. David W. Smith 33th Annual Workshop on Malformations and Morphogenesis. Lake Laniere, Georgia August, 2012

11. Be aware of the Dragon: Hearing loss from detection to diagnosis. Ball State University – Winter Series, February 22, 2013

12. Pharmacogenetics. Annual Meeting Pediatric Pharmacy Advocacy Group. Indianapolis, Indiana May 1st, 2013.

13. The New Genetics: Concepts we all should know. Indiana Speech, Language, and Hearing

Association annual meeting, September 21st, 2013.

14. The genetics of Ichtyosis. Family First. Annual Conference, Indianapolis, Indiana, June 2014

15. Mantaining the Health of Down syndrome Children. Down syndrome national congress. Indianapolis, Indiana July 2014

16. Genetics of Hearing loss: Cross Roads Conference on Hearing, Prudue University Nov, 2016, Lafayette, In

17. Hearing and Genetics, American Academy of Audiology annual meeting, April 2017, Indianapolis, In

RECENT CONTINUING EDUCATION COURSES:

David W. Smith 31th Annual Workshop on Malformations and Morphogenesis. Lake Arrow Head, California August, 2011

David W. Smith 32th Annual Workshop on Malformations and Morphogenesis. Lake Laniere, Georgia August, 2012

Annual American College of Medical Genetics Meeting, Phoenix, AZ, March 24-28, 20013

Annual American College of Medical Genetics Meeting, Nashville, TN, March 2014

David W. Smith 33th Annual Workshop on Malformations and Morphogenesis, The Fluno Center, University of Winsconsin, Madison Wi. July 2014.

American Society of Human Genetics, Baltimore MD, October 2015

David W. Smith 34th Annual Workshop on Malformations and Morphogenesis. Lake Arrow Head, California August, 2016

Annual American College of Medical Genetics Meeting, Phoenix, AZ, March 25-29, 20017

David W. Smith 34th Annual Workshop on Malformations and Morphogenesis. Stowe flake, Vermont August, 25-30, 2017

Early Hearing Detection & Intervention Annual Meeting, Denver, Colorado, March 18-20, 2018