CURRICULUM VITAE

Lori A. Sadler Sullivan, Ph.D.

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Business Address

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Personal

Date of birth: February 6, 1963. Citizenship: USA.

Education

- B.A. May 1984, Johns Hopkins University, Baltimore, Maryland. Major: Biology
- Ph.D. June 1990, Biology Department, University of California, Los Angeles
  Research Specialization: Molecular evolution
  Thesis Advisor: Clifford F. Brunk, Ph.D.
- Postdoctoral Fellow, University of Texas Health Science Center - Houston, June 1990-April 1991. Sponsor: Wen-Hsiung Li, Ph.D.
- Postdoctoral Fellow, University of Texas Health Science Center - Houston, April 1991-Jan. 1995. Sponsor: Stephen P. Daiger, Ph.D.

Employment

- Laboratory Technician, Johns Hopkins School of Hygiene and Public Health, Division of Biophysics, 1984.
- Research Associate, Department of Biology, University of California, Los Angeles, 1984-1990.
- Assistant Professor of Biological Sciences, Human Genetics Center, University of Texas - Houston School of Public Health, 1995 to 2003.
- Laboratory Supervisor, Laboratory for the Molecular Diagnosis of Inherited Eye Disease, University of Texas – Houston, CLIA ID#45D0935007, 1996 to 2020.
- Faculty Associate, Human Genetics Center, University of Texas - Houston School of Public Health, Sept. 2003 to present.

Consulting

- AGTC (Applied Genetics Technology Corporation), 2019 - 2022
- 4DMT (4D Molecular Therapeutics), 2021 - present

Honors and Professional Activities

- NIH Predoctoral Fellowship in Genetics and Regulatory Mechanisms, 1986-1989 (USPHS-GM Research Service Award).
- NIH Postdoctoral Fellowship, 1992-1994, "Candidate Genes for Retinal Degeneration on Chromosome 8".
- Chair, UT-Houston Committee on the Status of Women, 1999-2000.
- Coordinator, Clinical Genome Resource (ClinGen) Leber Congenital Amaurosis/ early onset Retinal Dystrophy Variant Curation Expert Panel, 2022 – present
- Member, Clinical Genome Resource (ClinGen) Retina Gene Curation Expert Panel, 2021 - present
- Member, Clinical Genome Resource (ClinGen) X-linked Inherited Retinal Disease Variant Curation Expert Panel, 2021 – present
- Member, Clinical Genome Resource (ClinGen) Education, Coordination and Training Working Group, 2022-present
- Member, NIH/NEI eyeGENE Expert Panel, 2024 – present
- Member, Clinical Genome Resource (ClinGen) Maculopathy Variant Curation Expert Panel, 2024 – present

Professional Memberships

- American Society of Human Genetics since 1993.
- Association for Research in Vision and Ophthalmology since 1991.
- University of Texas Association of Women Faculty 1993-1998.

Recent Major Research Support

Agency, title, period: National Eye Institute, NIH, “DNA Linkage Studies of Degenerative Retinal Diseases with Supplement”, 09/15/2008 - 08/31/2018
PI and role: Dr. Sullivan, Coinvestigator, (Dr. S.P. Daiger, PI)
Description: This project involves analysis of a large adRP cohort to identify genes and mutations causing autosomal dominant retinitis pigmentosa using classic methods such as linkage analysis as well as modern methods such as next-generation DNA sequencing.

Agency, title, period: National Eye Institute, NIH, “EyeGENE National Ophthalmic Disease Genotyping Network, Houston Subcontract”, 10/01/2007 - 10/31/2018
PI and role: Dr. Sullivan, Coinvestigator, (Dr. S.P. Daiger, PI)
Description: This subcontract provides support for DNA testing of samples from patients with autosomal dominant retinitis pigmentosa and related diseases, as part of the national EyeGENE Consortium.

Agency, title, period: Foundation Fighting Blindness, “Identification of the Remaining Genes and Mutations Causing Autosomal Dominant Retinitis Pigmentosa (adRP)”, 12/1/2018-11/30/2023
PI and role: Dr. Sullivan, Coinvestigator, (Dr. S.P. Daiger, PI)

Description: In prior research supported by the Foundation Fighting Blindness we have enrolled and collected blood samples from over 1,500 families with a clinical diagnosis of adRP. This project focuses on families in which mutations have not yet been found and involves collaboration with several other laboratories using novel bioinformatic and laboratory approaches to disease gene identification.

Agency, title, period: Foundation Fighting Blindness, "Registry of IRD-associated variants and cases", 01/01/2020-12/31/2022
PI and role: Dr. Sullivan, Curator adRP (Dr. Johan T. den Dunnen Dr.. Frans P.M. Cremers , PIs)
Description: This project is to collect, assess and register non-syndromic IRD, Bardet Biedl and Usher syndrome associated variants, phenotypes and cases in gene specific LOVDs.

Agency, title, period: Regents of the University of California-San Francisco (RUCSF)/Fogarty International Center/NIH/National Eye Institute, "Expert curation of clinically significant variants in genes for early onset retinal degeneration", Houston Consortium, 07/01/2022-05/31/2025
PI and role: Dr. Sullivan, PI Houston Consortium (Dr. Jacque Duncan, PI, UCSF)
Description: This project is to continue the process of establishing a variant curation expert panel (VCEP) within the NIH-sponsored Clinical Genome Resource (ClinGen), to curate genetic variants in genes associated with LCA and other early-onset retinal diseases. Curated variants will be entered in the ClinVar public database and will enable accurate, consistent, high quality interpretation of genetic test results and improve patient care.

Agency, title, period: Foundation Fighting Blindness, "Management and Curation of the RetNet database", 6/15/2024 – 6/14/2027.
PI and role: Dr. Sullivan, Curator
Description: This project will update, curate, and manage the "RetNet" database, a resource for the international IRD research community. RetNet has tracked new and updated gene discoveries in the IRD field since 1990 and is now part of the Foundation Fighting Blindness.

Publications

1. LA Sadler and CF Brunk. Phylogenetic relations among *Tetrahymena* species determined by DNA sequence analysis. In UCLA Symposia on Molecular and Cellular Biology, Vol. 122, (eds. M Clegg and SJ O'Brien) pp. 245-252, 1989.

2. CF Brunk and LA Sadler. Characterization of the promoter region of *Tetrahymena* genes. Nucl Acids Res. 18:323-329, 1990.

3. CF Brunk, RW Kahn, LA Sadler. Phylogenetic relationships among *Tetrahymena* species determined using the polymerase chain reaction. J Mol Evol. 30:290-297, 1990.

4. W-H Li and LA Sadler. Low nucleotide diversity in man. Genetics 129:513-523, 1991.

5. LA Sadler, SH Blanton, SP Daiger. Dinucleotide repeat polymorphism at the human tissue plasminogen activator gene (PLAT). Nucl Acids Res. 19:6058, 1991.

6. SH Blanton, JR Heckenlively, AW Cottingham, J Friedman, LA Sadler, M Wagner, LH Friedman, SP Daiger. Linkage mapping of autosomal dominant retinitis pigmentosa (RP1) to the pericentric region of human chromosome 8. Genomics 11:857-869, 1991

7. W-H Li and LA Sadler. DNA variation in humans and its implications for human evolution. Oxford Surveys in Evolutionary Biology, Vol. 8, Oxford University Press, 1992.

8. LA Sadler and CF Brunk. Phylogenetic relationships and unusual diversity in histone H4 proteins within the *Tetrahymena pyriformis* complex. Mol Biol Evol. 9:70-84, 1992.

9. AW Cottingham, LA Sadler, SH Blanton, MJ Wagner, DE Wells, JR Heckenlively, SP Daiger. A tight linkage cluster, with two new RFLPs (D8S96 and D8S108), in the interval 8cen-q13. Nucl Acids Res. 20:1426, 1992.

10. J Tomfohrde, S Wood, J Schertzer, MJ Wagner, DE Wells, J Parrish, LA Sadler, SH Blanton, SP Daiger, Z Wang, PJ Wilkie, JL Weber. Human chromosome 8 linkage map based on short tandem repeat polymorphisms: effect of genotyping errors. Genomics 14:144-152, 1992.

11. LA Sadler, KL McNally, NS Govind, CF Brunk, RK Trench. The nucleotide sequence of the small subunit ribosomal RNA gene from *Simbiodinium pilosum*, a symbiotic dinoflagellate. Curr Genet. 21:409-416, 1992.

12. T Steinbrueck, C Read, SP Daiger, LA Sadler, JL Weber, S Wood, H Donis-Keller. Chromosome 8. Science, 258:71-ff, 1992.

13. Y Wang, L Sadler, JT Hecht. Polymorphic dinucleotide repeat in a cartilage matrix protein (CRTM) gene. Hum Mol Genet. 1:780, 1993.

14. J Gu, L Sadler, S Daiger, D Wells, M Wagner. Dinucleotide repeat polymorphism at the CRH gene. Hum Mol Genet. 2:85, 1993.

15. S Wood, KB Othmane, USR Bergerheim, SH Blanton, R Bookstein, RA Clarke, SP Daiger, H Donis-Keller, D Drayna, S Kumar, RJ Leach, H-J Ludecke, J Oshima, LA Sadler, NK Spurr, T Steinbrueck, J Trapman, M Wagner, Z Wang, D Wells, CA Westbrook. Report of the first international workshop on human chromosome 8 mapping. Cytogenet Cell Genet. 64:133-146, 1993.

16. LS Sullivan, J Parrish, MJ Wagner, D Wells, SH Blanton, SP Daiger. Tetranucleotide repeat polymorphism (D8S582) for human EST00680. Hum Mol Genet. 3:386, 1994.

17. RE McGuire, AM Gannon, LS Sullivan, JA Rodriguez, SP Daiger. A second family with autosomal dominant retinitis pigmentosa linked to chromosome 7q (RP10): Evidence for a major adRP gene. Human Genetics 95:71-74, 1995.

18. SP Daiger, LA Sadler, JA Rodriguez. Correlation of phenotype with genotype in inherited retinal degeneration. Behavioral and Brain Sciences 18:491-506, 1995.

19. RE McGuire, LS Sullivan, SH Blanton, MW Church, JR Heckenlively, SP Daiger. X-linked dominant cone-rod degeneration: Linkage mapping of a new locus (RP15) to Xp22.13-p22.11. Am J Hum Genet 57:87-94, 1995

20. NK Spurr, S Blanton, R Bookstein, R Clarke, R Cottingham, S Daiger, D Drayna, P Faber, S Horrigan, K Kas, C Kirchgessner, S Kumar, RJ Leach, H-J Lüedecke, Y Nakamura, M-J Pébusque, S Ranta, E Sim, LS Sullivan, L Takle, J Vance, M Wagner, D Wells, C Westbrook, L Yaremko, D Zaletayev, O Zuffardi, S Wood. Report of the second international workshop on human chromosome 8 mapping 1994. Cytogenet. Cell Genet. 68:147-164, 1995.

21. LS Sullivan and SP Daiger. Inherited retinal degeneration: exceptional genetic and clinical heterogeneity. Molecular Medicine Today 2:380-386, 1996.

22. S Xu, M Denton, L Sullivan, SP Daiger, A Gal. Genetic mapping of RP1 on 8q11-q21 in an Australian family with autosomal dominant retinitis pigmentosa reduces the critical region to 4 cM between D8S601 and D8S285. Hum Genet 98:741-743, 1996.

23. MM Sohocki, LS Sullivan, WR Harrison, EJ Sodergren, F Elder, G Weinstock, S Tanase, SP Daiger. Molecular characterization and localization to 8q34 of the human glutamate pyruvate transaminase (GPT) locus. Genomics 40:247-252, 1997.

24. MM Sohocki, LS Sullivan, HA Mintz-Hittner, K Small, RE Ferrell, SP Daiger. Exclusion of atypical vitelliform macular dystrophy from 8q24.3 and from other known macular degenerative loci. Am J Hum Genet 61:239-241, 1997.

25. RW Yee, LS Sullivan, HT Lai, EL Stock, Y Lu, MN Khan, SH Blanton, SP Daiger. Linkage Mapping of Thiel-Behnke corneal dystrophy (CDB2) to chromosome 10q23-q24. Genomics, 46:152-154, 1997.

26. SP Daiger, RE McGuire, LS Sullivan, MM Sohocki, SH Blanton, P Humphries, ED Green, H Mintz-Hittner, JR Heckenlively. Progress in positional cloning of RP10 (7q31.3), RP1 (8q11-q21) and VMD1 (8q24). In "Degenerative Retinal Diseases", M LaVail, JG Hollyfield, RE Anderson, Eds, Plenum Publishing Co., pp277-289, 1997.

27. MM Sohocki, LS Sullivan, HA Mintz-Hittner, D Birch, JR Heckenlively, CL Freund, RR McInnes, SP Daiger. Cone-rod dystrophy, retinitis pigmentosa and dominant Leber congenital amaurosis associated with mutations in CRX, a photoreceptor transcription factor gene. Am. J. Hum Genet. 63:1307-1315, 1998.

28. MM Sohocki, KA Malone, LS Sullivan, SP Daiger. Localization of retina/pineal-expressed sequences: Identification of novel candidate genes for inherited retinal disorders. Genomics, 58:29-33, 1999.

29. K Malone, MM Sohocki, LS Sullivan, SP Daiger. Identifying and mapping novel retina-expressed ESTs from humans. Mol Vis.5:5, 1999 (http://www.molvis.org/molvis/v5/p5/)

30. LS Sullivan, JR Heckenlively, SJ Bowne, J Zuo, WA Hide, A Gal, M Denton, CF Inglehearn, SH Blanton, SP Daiger. Mutations in a novel retina-specific gene cause autosomal dominant retinitis pigmentosa. Nature Genet. 22:255-259, 1999.

31. SJ Bowne, SP Daiger, MM Hims, MM Sohocki, KA Malone, AB McKie, JR Heckenlively, DG Birch, CF Inglehearn, SS Bhattacharya, A Bird, LS Sullivan. Mutations in the RP1 gene causing autosomal dominant retinitis pigmentosa. Hum Mol Genet. 8:2121-2128, 1999.

32. MM Sohocki, SJ Bowne, LS Sullivan, S Blackshaw, CL Cepko, AM Payne, SS Bhattacharya, S Khaliq, S Qasim Mehdi, DG Birch, WR Harrison, FF Elder, JR Heckenlively, SP Daiger. Mutations in a new photoreceptor-pineal gene on 17p cause Leber congenital amaurosis. Nature Genet. 24:79-83, 2000.

33. SJ Bowne, LS Sullivan, L Ding, E Traer, SM Prescott, DG Birch, JR Heckenlively, A Kennan, P Humphries, SP Daiger. Evaluation of human diacylglycerol kinase iota, a homolog of *Drosophila rdgA*, in inherited retinopathy mapping to 7q. Mol Vis. 6:6-9, 2000.

34. MM Sohocki, SP Daiger, SJ Bowne, J Rodriguez, H Northrup, JR Heckenlively, DG Birch, H Mintz-Hittner, R Ruiz, R Lewis, D Saperstein, LS Sullivan. Prevalence of mutations causing retinitis pigmentosa and other inherited retinopathies. Hum Mut. 17:42-51, 2001.

35. MM Sohocki, LS Sullivan, DL Tirpak, SP Daiger. Comparative analysis of aryl-hydrocarbon receptor interacting protein-like 1 (Aipl1), a gene associated with inherited retinal disease in humans. Mamm Genome. 12:566-568, 2001

36. Q Liu, J Zhou, SP Daiger, DB Farber, JR Heckenlively, LS Sullivan, J Smith, J Zuo, AH Milam, EA Pierce. Identification and subcellular localization of the RP1 protein in human and mouse photoreceptors. Invest Ophthalmol Vis Sci. 43:22-32, 2002.

37. SJ Bowne, LS Sullivan, SH Blanton, CL Cepko, S Blackshaw, DG Birch, D Hughbanks-Wheaton, JR Heckenlively, SP Daiger. Mutations in the inosine monophosphate dehydrogenase 1 gene (IMPDH1) cause the RP10 form of autosomal dominant retinitis pigmentosa. Hum Mol Genet 11: 547-558, 2002.

38. J Gao, K Cheon, S Nusinowitz, Q Liu, D Bei, K Atkins, A Azimi, SP Daiger, DB Farber, JR Heckenlively, EA Pierce, LS Sullivan, J Zuo. Progressive photoreceptor degeneration, outer segment dysplasia, and rhodopsin mislocalization in mice with targeted disruption of the retinitis pigmentosa-1 (Rp1) gene. Proc Natl Acad Sci U S A. 99:698-703, 2002.

39. SJ Bowne, SP Daiger, KA Malone, JR Heckenlively, A Kennan, P Humphries, D Hughbanks-Wheaton, DG Birch, Q Liu, EA Pierce, J Zuo, Q Huang, DD Donovan, LS Sullivan. Characterization of RP1L1, a highly polymorphic paralog of the retinitis pigmentosa 1 (RP1) gene. Mol Vis. 9:129-137, 2003.

40. SC Khani, AJ Karoukis, JE Young, R Ambasudhan, T Burch, R Stockton, RA Lewis, LS Sullivan, SP Daiger, E Reichel, R Ayyagari. Late-onset autosomal dominant macular dystrophy with

choroidal neovascularization and nonexudative maculopathy associated with mutation in the RDS gene. Invest Ophthalmol Vis Sci. 44:3570-3577, 2003.

41. LS Sullivan, X Zhao, SJ Bowne, X Xu, SP Daiger, SB Yee, RW Yee. Exclusion of the human collagen type XVII (COL17A1) gene as the cause of Thiel-Behnke corneal dystrophy (CDB2) on chromosome 10q23-q25. Curr Eye Res. 27:223-226, 2003.

42. P Kozma, DK Hughbanks-Wheaton, KG Locke, GE Fish, AI Gire, CJ Spellicy, LS Sullivan, SJ Bowne, SP Daiger, DG Birch. Phenotypic characterization of a large family with RP10 autosomal-dominant retinitis pigmentosa: an Asp226Asn mutation in the IMPDH1 gene. Am J Ophthalmol. 140:858-867, 2005.

43. SJ Bowne, LS Sullivan, SE Mortimer, L Hedstrom, J Zhu, CJ Spellicy, AI Gire, D Hughbanks-Wheaton, DG Birch, RA Lewis, JR Heckenlively, SP Daiger. Spectrum and frequency of mutations in IMPDH1 associated with autosomal dominant retinitis pigmentosa and leber congenital amaurosis. Invest Ophthalmol Vis. Sci. 47:34-42, 2006.

44. LS Sullivan, SJ Bowne, DG Birch, D Hughbanks-Wheaton, JR Heckenlively, RA Lewis, CA Garcia, RS Ruiz, SH Blanton, H Northrup, AI Gire, R Seaman, H Duzkale, CJ Spellicy, J Zhu, SP Shankar, SP Daiger. Prevalence of disease-causing mutations in families with autosomal dominant retinitis pigmentosa: a screen of known genes in 200 families. Invest Ophthalmol Vis Sci. 47:3052-3064, 2006.

45. SJ Bowne, Q Liu, LS Sullivan, J Zhu, CJ Spellicy, C Bowes Rickman, EA Pierce, SP Daiger. Why do mutations in the ubiquitously expressed housekeeping gene IMPDH1 cause retina-specific photoreceptor degeneration? Invest. Ophthalmol Vis Sci. 47:3754-3765, 2006.

46. LS Sullivan, SJ Bowne, CR Seaman, SH Blanton, RA Lewis, JR Heckenlively, DG Birch, D Hughbanks-Wheaton, SP Daiger. Genomic rearrangements of the PRPF31 gene account for 2.5% of autosomal dominant retinitis pigmentosa. Invest Ophthalmol Vis Sci. 47:4579-4588, 2006.

47. SP Daiger, SP Shankar, AB Schindler, LS Sullivan, SJ Bowne, TM King, EW Daw, EM Stone, JR Heckenlively. Genetic factors modifying clinical expression of autosomal dominant RP. Adv Exp Med Biol. 572:3-8, 2006.

48. SP Daiger, SJ Bowne, LS Sullivan. Perspective on genes and mutations causing retinitis pigmentosa. Arch Ophthalmol. 125:151-158, 2007.

49. LS Sullivan, EB Baylin, R Font, SP Daiger, JS Pepose, TE Clinch, H Nakamura, XC Zhao, RW Yee. A novel mutation of the Keratin 12 gene responsible for a severe phenotype of Meesmann's corneal dystrophy. Mol Vis. 13:975-980, 2007.

50. CJ Spellicy, SP Daiger, LS Sullivan, J Zhu, Q Liu, EA Pierce, SJ Bowne. Characterization of retinal inosine monophosphate dehydrogenase 1 in several mammalian species. Mol Vis. 13:1866-1872, 2007.

51. A Gire, LS Sullivan, SJ Bowne, DG Birch, D Hughbanks-Wheaton, JR Heckenlively, SP Daiger. The Gly56Arg mutation in NR2E3 accounts for 1-2% of autosomal dominant retinitis pigmentosa. Mol Vis. 13:1970-1975, 2007.

52. SP Daiger, LS Sullivan, AI Gire, DG Birch, JR Heckenlively, SJ Bowne. Mutations in known genes account for 58% of autosomal dominant retinitis pigmentosa (adRP). Adv Exp Med Biol. 613:203-209, 2008.

53. SJ Bowne, LS Sullivan, AI Gire, DG Birch, D Hughbanks-Wheaton, JR Heckenlively, SP Daiger. Mutations in the TOPORS gene cause 1% of autosomal dominant retinitis pigmentosa. Mol Vis. 14:922-927, 2008.

54. L Köhn, SJ Bowne, LS Sullivan, SP Daiger, MSI Burstedt, K Kadzhaev, O Sandgren, I Golovleva. Breakpoint characterization of a novel ~59 kb genomic deletion on 19q13.42 in autosomal dominant retinitis pigmentosa with incomplete penetrance. Euro J Hum Genet 17:651-655, 2008.

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56. T Yamashita, J Liu, J Gao, S LeNoue, C Wang, J Kaminoh, SJ Bowne, LS Sullivan, SP Daiger, K Zhang, MEC Fitzgerald, VJ Kefalov, J Zuo. Essential and synergistic roles of RP1 and RP1L1 in rod photoreceptor axoneme and retinitis pigmentosa. J Neurosci. 29:9748-9760, 2009.

57. C Zhao, DL Bellur, S Lu, F Zhao, MA Grassi, SJ Bowne, LS Sullivan, SP Daiger, LJ Chen, CP Pang, K Zhao, JP Staley, C Larsson. Autosomal-dominant retinitis pigmentosa caused by a mutation in SNRNP200, a gene required for unwinding of U4/U6 snRNAs. Am J Hum Genet. 85:617-627, 2009.

58. SP Daiger, LS Sullivan, SJ Bowne, DG Birch DG, JR Heckenlively, EA Pierce, GM Weinstock. Targeted high-throughput DNA sequencing for gene discovery in retinitis pigmentosa. Adv Exp Med Biol. 664:325-331, 2010.

59. CJ Spellicy, D Xu, G Cobb, L Hedstrom SJ Bowne, LS Sullivan, SP Daiger. Investigating the mechanism of disease in the RP10 form of retinitis pigmentosa. Adv Exp Med Biol. 664:541-548, 2010.

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61. L Köhn, Kohl, SJ Bowne, LS Sullivan, Kellner, SP Daiger, O Sandgren, I Golovleva. PITPNM3 is an uncommon cause of cone and cone-rod dystrophies. Ophthalmic Genetics, 31:139-140, 2010.

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63. AT Fahim, SJ Bowne, LS Sullivan, K Clark, JT Williams, DK Wheaton, DG Birch, SP Daiger. Allelic heterogeneity and genetic modifier loci contribute to clinical variation in males with X-linked retinitis pigmentosa due to mutations in RPGR. PLoS One, 6:e23021, 2011. PubMed PMID: 21857984.

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68. SJ Bowne , LS Sullivan, CE Avery, EM Sasser, A Roorda, JL Duncan, DH Wheaton, DG Birch, KE Branham, JR Heckenlively, PA Sieving , SP Daiger. Mutations in the small nuclear riboprotein 200 kDa gene (SNRNP200) cause 1.6% of autosomal dominant retinitis pigmentosa. Mol Vis. 19:2407-2417, 2013. PubMed PMID: 24319334.

69. LS Sullivan, SJ Bowne , MJ Reeves, D Blain, K Goetz, V Ndifor, S Vitez, X Wang, SJ Tumminia, SP Daiger. Prevalence of mutations in eyeGENE probands with a diagnosis of autosomal dominant retinitis pigmentosa. Invest Ophthalmol Vis Sci. 54:6255-6261, 2013. PubMed PMID: 23950152.

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Recent Abstracts

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