Brain Vascular Malformation Consortium

Book Chapters

1. Morrison L, Akers A. Cerebral Cavernous Malformation, Familial. In: Pagon RA, Adam MP, Bird TD, et al., eds. GeneReviews(R). Seattle WA: University of Washington, Seattle; 2003 (Updated 2011 May 31).

2. Kim H, Pawlikowska L, Young WL. Molecular and genetic aspects of brain vascular malformation. In: Mohr JP, Wolf PA, Grotta JC, Moskowitz MA, Mayberg M, Von Kummer R, eds. *Stroke: Pathophysiology, diagnosis, and Management*. Philadelphia: Churchill Livingstone Elsevier; 2010.

3. Kim H, Pawlikowska L, Young W. Genetics and vascular biology of brain vascular malformations. In: Mohr J, Wolf P, Grotta J, Moskowitz M, Mayberg M, vonKummer R, eds. Stroke: Pathophysiology, Diagnosis, and Management. Philadelphia, PA: Churchill Livingstone Elsevier; 2011:169-186.

4. Morrison L. Familial cavernous malformations: a historical survey In: Rigamonti D, ed. Cavernous Malformations of the Nervous System. Cambridge: Cambridge University Press; 2011:15-20.

5. Morrison L. Genetic counseling In: Rigamonti D, ed. Cavernous Malformations of the Nervous System. Cambridge: Cambridge University Press; 2011:181-184.

Abstracted Presented at Conferences

1. Khan Y, Hart B, Morrison L. CCM in Hispanic children in New Mexico Paper presented at: 2nd Conference on Clinical Research for Rare Diseases; September 21, 2010; Washington, DC.

2. Khan Y, Kim H, Hart B, Morrison L. CCM in Hispanic children in New Mexico. Paper presented at: 39th Annual Meeting of the Child Neurology Society; October 13-16, 2010; Providence, RI.

3. Morrison L. CCM in Hispanic children Paper presented at: 39th Annual Meeting of the Child Neurology Society; October 13-16, 2010; Providence, RI.

4. Gonzales N, Gonzales J, Baca B, Morrision L. Finding the founder of CCM1 in New Mexico. Paper presented at: 6th Annual Pathobiology of CCM Scientific Meeting; November 1-2, 2010; Washington, DC.

5. Morrison L. Clinical characteristics in CCM1 Paper presented at: 6th Annual Pathobiology of CCM Scientific Meeting; November 1-2, 2010; Washington, DC.

6. Khan Y, Kim H, Hart B, Morrison L. CCM in Hispanic children in New Mexico Paper presented at: 6th Annual Pathobiology of CCM Scientific Meeting; November 1-2, 2010; Washington, DC

7. Morrison L. CCM in Hispanic children Paper presented at: Radiologic Society of North America 96th Scientific Assembly and Annual Meeting; November 28 - December 3, 2010; Chicago, IL.

8. Choquet H, Nelson J, Pawlikowska L, et al. Clinical factors associated with lesion count in familial cerebral cavernous malformation type 1 patients with the common Hispanic mutation. Paper presented at: RDCRN 3rd Conference on Clinical Research for Rare Diseases; October 2, 2012; Washington, DC.

9. Lance E, Lanier K, A Z, Comi A. Stimulant use in patients with Sturge-Weber Syndrome: safety and efficacy. Paper presented at: Child Neurology Society 41st Annual Meeting; October 31 - November 3, 2012; Hungtington Beach, CA.

10. Sreenivasan A, Curatolo A, Connors S, Moses M, Comi A. Angiogenesis factors as urine biomarkers in Sturge-Weber Syndrome. Paper presented at: Child Neurology Society 41st Annual Meeting; October 31 - November 3, 2012; Huntington Beach, CA.

11. Choquet H, Nelson J, Pawlikowska L, et al. Clinical factors associated with lesion count in familial cerebral cavernous malformation type 1 patients with the common Hispanic mutation Paper presented at: 8th Annual CCM Scientific Meeting; November 15-16, 2012; Washington, DC.

12. Morrison L, Wegele A, Baca B. Cutaneous features of CCM1-CHM cohort Paper presented at: 8th Annual CCM Scientific Meeting; November 15-16, 2012; Washington, DC.

13. Baca B, Hart B, Kim H, Morrison L. Lesion burden, location, and clinical characteristics in a genetically unique cohort of patients with cerebral cavernous malformations. Paper presented at: Radiologic Society of North America 96th Scientific Assembly and Annual Meeting; November 25-30, 2012; Chicago, IL.

14. Choquet H, Pawlikowska L, Nelson J, et al. Association of variants in inflammatory genes with lesion burden in familial CCM1. Paper presented at: 10th HHT Scientific Conference; June 12-15, 2013; Cork, Ireland.

15. Morrison L, Hart B, Baca B, et al. A comparison of hemorrhage and nonhemorrhage in patients with CCM1 common Hispanic mutation Paper presented at: 10th HHT Scientific Conference; June 12-15, 2013; Cork, Ireland.

16. Choquet H, Pawlikowska L, Nelson J, et al. Association of variants in inflammatory genes with disease severity in familial cerebral cavernous malformations type 1. Paper presented at: American Society of Human Genetics 63rd Annual Meeting; October 22-26, 2013; Boston, MA.

17. Choquet H, Nelson J, Pawlikowska L, et al. Association of common variants in immune response genes with severity of familiar cerebral cavernous malformation type 1. Paper presented at: 9th Annual CCM Scientific Meeting; November 7-8, 2013; Washington, DC.

18. Morrison L, Hart B, Nelson J, et al. Predictors of functional outcome in patients with CCM1 common Hispanic mutation Paper presented at: 9th Annual CCM Scientific Meeting; November 7-8, 2013; Washington, DC.

Conference Proceedings

1. Gallione C, Qin Y, Chu P, Akers A, Young W, Marchuk D. Re-examination of the two-hit hypthesis for HHT pathogenesis Paper presented at: 8th International Hereditary Hemorrhagic Telangiectasia Scientific Conference; May 27-31, 2009; Spain.

2. Choquet H, Nelson J, Pawlikowska L, et al. Clinical factors associated with lesion count in familial cerebral cavernous malformation Type 1 patients with the common Hispanic mutation Paper presented at: International Stroke Conference; February 5-8, 2013; Honolulu, HI.

3. Gossage J, Kim H, Faughnan M, BrainVascularMalformationConsortium. Female sex and ENG mutation are associated with an increased risk of PAVM in patients with definite HHT Paper presented at: 10th International Hereditary Hemorrhagic Telangiectasia Scientific Conference; June 12-15, 2013; Cork, Ireland.

4. Latino G, Faughnan M, Kim H, Young W, BrainVascularMalformationConsortium. A hereditary hemorrhagic telangiectasia severity score Paper presented at: 10th International Hereditary Hemorrhagic Telangiectasia Scientific Conference; June 12-15, 2013; Cork, Ireland.

5. Lin D, Zessler A, Young W, Faughnan M, BrainVascularMalformationConsortium. Age of presentation in HHT: Brain AVM diagnosis vs. epistaxis Paper presented at: 10th International Hereditary Hemorrhagic Telangiectasia Scientific Conference; June 12-15, 2013; Cork, Ireland.

6. Montifar M, Kasthuri R, Kim H, Young W, BrainVascularMalformationConsortium. Anemia is an important clinical problem in HHT. Paper presented at: 10th International Hereditary Hemorrhagic Telangiectasia Scientific Conference; June 12-15, 2013; Cork, Ireland.

7. Nishida T, terBrugge K, Krings T, Henderson K, White R. Micro brain vascular malformations associated with hereditary hemorrhagic telangiectasia: arteriovenous malformations and capillary malformations. Paper presented at: 10th International Hereditary Hemorrhagic Telangiectasia Scientific Conference; June 12-15, 2013; Cork, Ireland.

8. terBrugge K, T N, Krings T. Coincidental and acquired neurovascular malformations and shunts associated with HHT disorder Paper presented at: 10th International Hereditary Hemorrhagic Telangiectasia Scientific Conference; June 12-15, 2013; Cork, Ireland.

9. Choquet H, Nelson J, Pawlikowska L, et al. Association of common variants in immune response genes with severity of familial cerebral cavernous malformation type 1. Paper presented at: International Stroke Conference February 12-14, 2014; San Diego, CA.

10. Kim H, Nelson J, Krings T, terBrugge K, Young W, Faughnan M. Hemorrhage rates from brain arteriovenous malformations in HHT patients. Paper presented at: International Stroke Conference February 12-14, 2014; San Diego, CA.

Journal Articles

1. Ewen JB, Kossoff EH, Crone NE, Lin DD, Lakshmanan BM, Ferenc LM, Comi AM. Use of quantitative EEG in infants with port-wine birthmark to assess for Sturge-Weber brain involvement. *Clin. Neurophysiol.* Aug 2009;120(8):1433-1440. PMID: 19589723, PMCID: PMC2754702
2. Kossoff EH, Ferenc L, Comi AM. An infantile-onset, severe, yet sporadic seizure pattern is common in Sturge-Weber syndrome. *Epilepsia.* Sep 2009;50(9):2154-2157. PMID: 19389148
3. Leblanc GG, Golanov E, Awad IA, Young WL. Biology of vascular malformations of the brain. *Stroke.* Dec 2009;40(12):e694-702. PMID: 19834013, PMCID: PMC2810509
4. Zabel TA, Reesman J, Wodka EL, Gray R, Suskauer SJ, Turin E, Ferenc LM, Lin DD, Kossoff EH, Comi AM. Neuropsychological features and risk factors in children with Sturge-Weber syndrome: four case reports. *Clin Neuropsychol.* 2010;24(5):841-859. PMID: 20560093
5. Petersen TA, Morrison LA, Schrader RM, Hart BL. Familial versus sporadic cavernous malformations: differences in developmental venous anomaly association and lesion phenotype. *AJNR Am. J. Neuroradiol.* Feb 2010;31(2):377-382. PMID: 19833796
6. Arulrajah S, Ertan G, A MC, Tekes A, Lin DL, Huisman TA. MRI with diffusion-weighted imaging in children and young adults with simultaneous supra- and infratentorial manifestations of Sturge-Weber syndrome. *J. Neuroradiol.* Mar 2010;37(1):51-59. PMID: 19570579
7. Suskauer SJ, Trovato MK, Zabel TA, Comi AM. Physiatric findings in individuals with Sturge-Weber syndrome. *Am. J. Phys. Med. Rehabil.* Apr 2010;89(4):323-330. PMID: 20068437, PMCID: PMC3189450
8. Bharatha A, Faughnan ME, Kim H, Pourmohamad T, Krings T, Bayrak-Toydemir P, Pawlikowska L, McCulloch CE, Lawton MT, Dowd CF, Young WL, Terbrugge KG. Brain arteriovenous malformation multiplicity predicts the diagnosis of hereditary hemorrhagic telangiectasia: quantitative assessment. *Stroke.* Jan 2012;43(1):72-78. PMID: 22034007, PMCID: PMC3727386
9. Lo W, Marchuk DA, Ball KL, Juhasz C, Jordan LC, Ewen JB, Comi A. Updates and future horizons on the understanding, diagnosis, and treatment of Sturge-Weber syndrome brain involvement. *Dev. Med. Child Neurol.* Mar 2012;54(3):214-223. PMID: 22191476, PMCID: PMC3805257
10. Kadam SD, Gucek M, Cole RN, Watkins PA, Comi AM. Cell proliferation and oxidative stress pathways are modified in fibroblasts from Sturge-Weber syndrome patients. *Arch. Dermatol. Res.* Apr 2012;304(3):229-235. PMID: 22402795
11. Lance EI, Sreenivasan AK, Zabel TA, Kossoff EH, Comi AM. Aspirin use in Sturge-Weber syndrome: side effects and clinical outcomes. *J. Child Neurol.* Feb 2013;28(2):213-218. PMID: 23112247
12. Nishida T, Faughnan ME, Krings T, Chakinala M, Gossage JR, Young WL, Kim H, Pourmohamad T, Henderson KJ, Schrum SD, James M, Quinnine N, Bharatha A, Terbrugge KG, White RI, Jr. Brain arteriovenous malformations associated with hereditary hemorrhagic telangiectasia: gene-phenotype correlations. *Am. J. Med. Genet. A.* Nov 2012;158A(11):2829-2834. PMID: 22991266, PMCID: PMC3610331
13. Siddique L, Sreenivasan A, Comi AM, Germain-Lee EL. Importance of utilizing a sensitive free thyroxine assay in Sturge-Weber syndrome. *J. Child Neurol.* Feb 2013;28(2):269-274. PMID: 23112245

14. Akers A, Ball KL, Clancy M, et al. Brain Vascular Malformation Consortium: overview, progress, and future directions. *The Journal of Rare Disorders*. April 2013;1(1):1-15. PMID: 25221778, PMCID: PMC4160161

15. Lopez J, Yeom KW, Comi A, Van Haren K. Case report of subdural hematoma in a patient with Sturge-Weber syndrome and literature review: questions and implications for therapy. *J. Child Neurol.* May 2013;28(5):672-675. PMID: 22805242

16. Shirley MD, Tang H, Gallione CJ, Baugher JD, Frelin LP, Cohen B, North PE, Marchuk DA, Comi AM, Pevsner J. Sturge-Weber syndrome and port-wine stains caused by somatic mutation in GNAQ. *N. Engl. J. Med.* May 23 2013;368(21):1971-1979. PMID: 23656586, PMCID: PMC3749068

17. Sreenivasan AK, Bachur CD, Lanier KE, et al. Urine vascular biomarkers in Sturge-Weber syndrome. *Vasc. Med.* Jun 2013;18(3):122-128. PMID: 23720035

18. Vlachou PA, Colak E, Koculym A, et al. Improvement of ischemic cholangiopathy in three patients with hereditary hemorrhagic telangiectasia following treatment with bevacizumab. *J. Hepatol.* Jul 2013;59(1):186-189. PMID: 23439260

19. Arora KS, Quigley HA, Comi AM, Miller RB, Jampel HD. Increased choroidal thickness in patients with Sturge-Weber syndrome. *JAMA ophthalmology.* Sep 2013;131(9):1216-1219. PMID: 23828561

20. Bachur CD, Comi AM. Sturge-weber syndrome. *Curr. Treat. Options Neurol.* Oct 2013;15(5):607-617. PMID: 23907585

21. Hart BL, Taheri S, Rosenberg GA, Morrison LA. Dynamic contrast-enhanced MRI evaluation of cerebral cavernous malformations. *Translational stroke research*. Oct 2013;4(5):500-506. PMID: 24323376, PMCID: PMC3939060

22. Comi AM, Marchuk DA, Pevsner J. A needle in a haystack: sturge-weber syndrome gene discovery. *Pediatr. Neurol.* Dec 2013;49(6):391-392. PMID: 24075845

23. Choquet H, Nelson J, Pawlikowska L, et al. Association of cardiovascular risk factors with disease severity in cerebral cavernous malformation type 1 subjects with the common Hispanic mutation. *Cerebrovasc. Dis.* 2014;37(1):57-63. PMID: 24401931, PMCID: PMC3995158

24. Choquet H, Pawlikowska L, Nelson J, et al. Polymorphisms in inflammatory and immune response genes associated with cerebral cavernous malformation type 1 severity. *Cerebrovasc. Dis.* 2014;38(6):433-440. PMID: 25472749, PMCID: PMC4297571

25. Cheng KH, Mariampillai A, Lee KK, et al. Histogram flow mapping with optical coherence tomography for in vivo skin angiography of hereditary hemorrhagic telangiectasia. *J. Biomed. Opt.* Aug 2014;19(8):086015. PMID: 25140883, PMC4407667

26. Latino GA, Kim H, Nelson J, Pawlikowska L, Young W, Faughnan ME. Severity score for hereditary hemorrhagic telangiectasia. *Orphanet J. Rare Dis.* 2014;9:188. PMID: 25928712, PMCID: PMC4302697

27. Lance EI, Lanier KE, Zabel TA, Comi AM. Stimulant use in patients with sturge-weber syndrome: safety and efficacy. *Pediatr. Neurol.* Nov 2014;51(5):675-680. PMID: 25439578, PMCID: PMC4392725

28. Reidy TG, Suskauer SJ, Bachur CD, McCulloch CE, Comi AM. Preliminary reliability and validity of a battery for assessing functional skills in children with Sturge-Weber syndrome. *Childs Nerv. Syst.* Dec 2014;30(12):2027-2036. PMID: 25344741, PMCID: PMC4276129

29. Choquet H, Pawlikowska L, Lawton MT, Kim H. Genetics of cerebral cavernous malformations: current status and future prospects. *J. Neurosurg. Sci.* 2015;59(3):211-220. PMID: 25900426, PMCID: PMC4461471

30. Comi A. Current Therapeutic Options in Sturge-Weber Syndrome. *Semin Pediatr Neurol.* 2015;22(4):295-301. PMID: 26706016, PMCID: PMC4943027

31. Golden M, Saeidi S, Liem B, Marchand E, Morrison L, Hart B. Sensitivity of patients with familial cerebral cavernous malformations to therapeutic radiation. *J. Med. Imaging Radiat. Oncol.* Feb 2015;59(1):134-136. PMID: 25565562, PMCID: PMC4437719

32. Golden MJ, Morrison LA, Kim H, Hart BL. Increased number of white matter lesions in patients with familial cerebral cavernous malformations. *AJNR Am. J. Neuroradiol.* May 2015;36(5):899-903. PMID: 25556204, PMCID: PMC4433814

33. Kim H, Nelson J, Krings T, et al. Hemorrhage rates from brain arteriovenous malformation in patients with hereditary hemorrhagic telangiectasia. *Stroke.* May 2015;46(5):1362-1364. PMID: 25858236, PMCID: PMC4415515

34. Krings T, Kim H, Power S, et al. Neurovascular manifestations in hereditary hemorrhagic telangiectasia: imaging features and genotype-phenotype correlations. *AJNR Am. J. Neuroradiol.* May 2015;36(5):863-870. PMID: 25572952, PMCID: PMC4433843

35. Kavanaugh B, Sreenivasan A, Bachur C, Papazoglou A, Comi A, Zabel TA. Intellectual and adaptive functioning in Sturge-Weber Syndrome. *Child neuropsychology : a journal on normal and abnormal development in childhood and adolescence.* May 8 2015:1-14. PMID: 25952468, PMCID: PMC4868126

36. Pawlikowska L, Nelson J, Guo DE, et al. The ACVRL1 c.314-35A>G polymorphism is associated with organ vascular malformations in hereditary hemorrhagic telangiectasia patients with ENG mutations, but not in patients with ACVRL1 mutations. *Am. J. Med. Genet. A.* Jun 2015;167(6):1262-1267. PMID: 25847705, PMCID: PMC4449292

37. Choquet H, Trapani E, Goitre L, et al. Cytochrome P450 and matrix metalloproteinase genetic modifiers of disease severity in Cerebral Cavernous Malformation type 1. *Free Radic Biol Med.* 2016;92:100-109. PMID: 26795600, PMCID: PMC4774945

38. Comi AM, Sahin M, Hammill A, et al. Leveraging a Sturge-Weber Gene Discovery: An Agenda for Future Research. *Pediatr Neurol.* 2016;58:12-24. PMID: 27268758, PMCID: PMC5509161

39. Merkel PA, Manion M, Gopal-Srivastava R, et al. The partnership of patient advocacy groups and clinical investigators in the rare diseases clinical research network. *Orphanet J. Rare Dis.* 2016;11(1):66. PMID: 27194034, PMCID: PMC4870759

40. Walcott BP, Choudhri O, Lawton MT. Brainstem cavernous malformations: Natural history versus surgical management. *J. Clin. Neurosci.* 2016. PMID: 27320373, PMCID: PMC5011020

41. Walcott BP, Winkler EA, Rouleau GA, Lawton MT. Molecular, Cellular, and Genetic Determinants of Sporadic Brain Arteriovenous Malformations. *Neurosurgery.* 2016;63 Suppl 1:37-42. PMID: 27399362, PMCID: PMC4941635

42. Kaplan EH, Kossoff EH, Bachur CD, et al. Anticonvulsant Efficacy in Sturge-Weber Syndrome. *Pediatr. Neurol.* Jan 11 2016. PMID: 26997037

43. Lo WD, Kumar R. Arterial Ischemic Stroke in Children and Young Adults. *Continuum (Minneapolis, Minn).* 2017;23(1, Cerebrovascular Disease):158-180. PMID: 28157749

44. Meybodi AT, Kim H, Nelson J, et al. Surgical Treatment vs Nonsurgical Treatment for Brain Arteriovenous Malformations in Patients with Hereditary Hemorrhagic Telangiectasia: A Retrospective Multicenter Consortium Study. *Neurosurgery.* 2017. PMID: 28973426

45. Strickland CD, Eberhardt SC, Bartlett MR, et al. Familial Cerebral Cavernous Malformations Are Associated with Adrenal Calcifications on CT Scans: An Imaging Biomarker for a Hereditary Cerebrovascular Condition. *Radiology.* 2017;284(2):443-450. PMID: 28318403, PMCID: PMC5519414

46. Zou X, Hart BL, Mabray M, et al. Automated algorithm for counting microbleeds in patients with familial cerebral cavernous malformations. *Neuroradiology.* 2017;59(7):685-690. PMID: 28534135, PMCID: PMC5501247