Curriculum Vitae

Personal information

Miikka Vikkula

Work experience

Since 1 Oct 2013 Full Professor of Human Genetics at Université catholique de Louvain

Since 1 Oct 2008 Honorary Maître de recherche, FNRS, Belgium

Since 15 Sept 2004 Member of the Directorate of de Duve Institute, Brussels, Belgium

Since 14 Jan 2004 Member of de Duve Institute, Brussels, Belgium

Oct'11 – Sept'13 Coordinator of the Centre for Human Genetics, CUSL, Brussels, Belgium

Oct'08 – Sept'13 Professor of Human Genetics, Department of biochemistry and cellular biology, Faculté de médecine, Université catholique de Louvain

Oct'04-Sept'08 Maître de Recherche, FNRS, Brussels, Belgium (Associate professor)

Oct'05-Sept'08 Chargé de cours définitif à temps partiel, Department of biochemistry and cellular biology, Faculté de médecine, Université catholique de Louvain

Oct'00-Sept'05 Chargé de cours temporaire à temps partiel, UCL, Fac. de médecine, Dept. of biochemistry and cellular biology

Oct'00-Sep'04 Chercheur qualifié, FNRS (Assistant professor)

Apr'97-Jan'04 Associate Member of de Duve Institute, Brussels, Belgium.

Feb'97-Sept'00 Guest Investigator, de Duve Institute, Brussels, Belgium.

Sep'93-Jan'97 Research Associate, Department of Cell Biology, Harvard Medical School, Boston, MA, USA. (PI: Professor Bjorn R Olsen)

Jun'93-Aug'93 Research Associate, National Public Health Institute, Helsinki, Finland. (PI: Professor Leena Peltonen)

Mar'93-May'93 Internship in internal medicine, District Hospital of Porvoo, Finland.

May'87-Feb'93 MD/PhD student in Department of Human Molecular Genetics, National Public Health Institute, Helsinki, Finland. (Supervisor: Professor Leena Peltonen)

May'89-Aug'89+ Visiting MD/PhD student in Department of Biochemistry and

Jun'88-Jul' 88 Molecular Biology, Thomas Jefferson University, Philadelphia, USA.

Education and training

2019-Ministry of Health, Brussels, Belgium
Specialist in clinical genetics

2000-Université catholique de Louvain, Brussels, Belgium Agrégé de l'Enseignement Supérieur (Molecular Genetics)(Ph.D.)

1993-University of Helsinki, Helsinki, Finland
Licensed M.D. (General Medicine)

1993-University of Helsinki, Helsinki, Finland
Ph.D. (Molecular Genetics)

1992-University of Helsinki, Helsinki, Finland
M.D. (Medicine)

1984-Pohjois-Tapiola High School, Helsinki, Finland
Baccalaureate

Additional information

Publications

Vikkula M, Peltonen L. Structural analyses of the polymorphic area in type II collagen gene. FEBS Lett. Jul 3 1989;250(2):171-4. IF=3.361
Vikkula M, Metsaranta M, Syvanen AC, Ala-Kokko L, Vuorio E, Peltonen L. Structural analysis of the regulatory elements of the type-II procollagen gene. Conservation of promoter and first intron sequences between human and mouse. Biochem J. Jul 1 1992;285 (Pt 1):287-94. IF=3.716
Vikkula M, Palotie A, Ritvaniemi P, Ott J, Ala-Kokko L, Sievers U, Aho K, Peltonen L. Early-onset osteoarthritis linked to the type II procollagen gene. Detailed clinical phenotype and further analyses of the gene. Arthritis Rheum. Mar 1993;36(3):401-9. IF=5,502
Vikkula M, Nissila M, Hirvensalo E, Nuotio P, Palotie A, Aho K, Peltonen L. Multiallelic polymorphism of the cartilage collagen gene: no association with osteoarthrosis. Ann Rheum Dis. Oct 1993;52(10):762-4. IF=1.630
Vikkula M, Ritvaniemi P, Vuorio AF, Kaitila I, Ala-Kokko L, Peltonen L. A mutation in the amino-terminal end of the triple helix of type II collagen causing severe osteochondrodysplasia. Genomics 1993, 16(1) :282-285. IF=5.433
Vikkula M, Metsäranta m, Ala-Kokko L. Type II collagen mutations in rare and common cartilage diseases. Ann Med 1994, 26(2) :107-114. IF=1.129
Vikkula M, Mariman EC, Lui VCH, Zhidkova NI, Tiller GE, Goldring M, van Beersum SEC, de Waal Malefijt MC, van den Hoogen FHJ, Ropers HH, Mayne R, Cheah KSE, Olsen BR, Warman ML, Brunner HG. Autosomal dominant and recessive osteochondrodysplasias associated with the COL11A2 locus. Cell. Feb 10 1995;80(3):431-7. IF=40.481
Vikkula M, Boon LM, Carraway III KL, Calvert JT, Diamonti AJ, Goumnerov B, Pasyk KA, Marchuk DA, Warman ML, Cantley LC, Mulliken JB, Olsen BR. Vascular dysmorphogenesis caused by an activating mutation in the receptor tyrosine kinase TIE2. Cell. Dec 27 1996;87(7):1181-90. IF=40.997
Jüppner H, Schipani E, Bastepe M, Cole DE, Lawson ML, Mannstadt M, Hendy GN, Plotkin H, Koshiyama h, Koh T, Crawford JD, Olsen BR, Vikkula M. The gene responsible for pseudohypoparathyroidism type Ib is paternally imprinted and maps in four unrelated kindreds to chromosome 20q13.3. Proc Natl Acad Sci USA. Sep 29 1998;95(20):11798-803. IF=10.426
Boon LM, Brouillard P, Irrthum A, Karttunen L, Warman ML, Rudolph R, Mulliken JB, Olsen BR, Vikkula M. A gene for inherited cutaneous venous anomalies ("glomangiomas") localizes to chromosome 1p21-22. Am J Hum Genet. Jul 1999;65(1):125-133. IF=10.426
Brouillard P, Olsen BR, Vikkula M. High-resolution physical and transcript map of the locus for venous malformations with glomus cells (VMGLOM) on chromosome 1p21-p22. Genomics. Jul 1 2000;67(1):96-101. IF=3.425
Eerola I, Plate KH, Spiegel R, Boon LM, Mulliken JB, Vikkula M. KRIT1 is mutated in hyperkeratotic cutaneous capillary-venous malformation associated with cerebral capillary malformation. Hum Mol Genet. May 22 2000;9(9):1351-5. IF=9.048
Irrthum A, Karkkainen MJ, Devriendt K, Alitalo K, Vikkula M. Congenital hereditary lymphedema caused by a mutation that inactivates VEGFR3 tyrosine kinase. Am J Hum Genet. Aug 2000;67(2):295-301. IF=10.351
Irrthum A, Brouillard P, Enjolras O, Gibbs NF, Eichenfeld LF, Olsen BR, Mulliken JB, Boon LM, Vikkula M. Linkage disequilibrium narrows locus for venous malformation with glomus cells (VMGLOM) to a single 1.48 Mbp YAC. Eur J Hum Genet. Jan 2001;9(1):34-8. IF=3.173
Eerola I, McIntyre B, Vikkula M. Identification of eight novel 5'-exons in cerebral capillary malformation gene-1 (CCM1) encoding KRIT1. Biochim Biophys Acta – Gene Struct Expr Feb 16 2001;1517(3):464-7. IF=3.536
Brouillard P, Boon LM, Mulliken JB, Ghassibé M, Warman ML, Tan OT, Olsen BR, Vikkula M. Mutations in a novel factor, glomulin, are responsible for glomuvenous malformations ("glomangiomas"). Am J Hum Genet. Apr 2002;70(4):866-74. IF=10.649
Eerola I, Boon LM, Watanabe S, Grynberg H, Mulliken JB, Vikkula M. Locus for susceptibility for familial capillary malformation ('port-wine stain') maps to 5q. Eur J Hum Genet. Jun 2002;10(6):375-380. IF=3.136
Gutierrez-Roelens I, Sluysmans T, Gewillig M, Devriendt K, Vikkula M. Progressive AV-block and anomalous venous return among cardiac anomalies associated with two novel missense mutations in the CSX/NKX2-5 gene. Hum Mutat. Jul 2002;20(1):75-6. IF=6.894
Renard L, Godfraind C, Boon LM, Vikkula M. A novel mutation in the SDHD gene in a family with inherited paragangliomas--implications of genetic diagnosis for follow up and treatment. Head Neck. Feb 2003;25(2):146-51. IF=1.805
Irrthum A, Devriendt K, Chitayat D, Matthijs G, Glade C, Steijlen PM, Fryns J-P, Van Steensen MAM, Vikkula M. Mutations in the transcription factor gene SOX18 underlie recessive and dominant forms of hypotrichosis-lymphedema-telangiectasia. Am J Hum Genet. Jun 2003;72(6):1470-78. IF=11.602
Godfraind C, Rousseau E, Ruchoux MM, Scaravilli F, Vikkula M. Tumour necrosis and microvascular proliferation are associated with 9p deletion and CDKN2A alterations in 1p/19q-deleted oligodendrogliomas. Neuropathol Appl Neurobiol. Oct 2003;29(5):462-71. IF=3.022
Ghassibe M, Revencu N, Bayet B, Gillerot Y, Vanwijck R, Veellen-Dumoulin C, Vikkula M. Gene symbol: IRF6. Disease: Van der Woude syndrome. Hum Genet. Nov 2003;113(6):556-8. IF=4.022;
Eerola I, Boon LM, Mulliken JB, Burrows PE, Vanwijck R, Vikkula M. Capillary malformation-arteriovenous malformation, a new clinical and genetic disorder caused by RASA1 mutations. Am J Hum Genet. Dec 2003;73(6):1240-9. IF=11.602
Rousseau E, Ruchoux MM, Scaravilli F, Chapon F, Vinchon M, De Smet C, Godfraind C, Vikkula M. CDKN2A, CDKN2B and p14ARF are frequently and differentially methylated in ependymal tumours. Neuropathol Appl Neurobiol. Dec 2003;29(6):574-83. IF=3.022
Ghassibe M, Revencu N, Bayet B, Gillerot Y, Vanwijck R, Veellen-Dumoulin C, Vikkula M. Six families with van der Woude and/or popliteal pterygium syndrome: all with a mutation in the IRF6 gene. J Med Genet. Feb 2004;41(2):e15. IF=4.112
McIntyre BA, Brouillard P, Aerts V, Gutierrez-Roelens I, Vikkula M. Glomulin is predominantly expressed in vascular smooth muscle cells in the embryonic and adult mouse. Gene Expr Patterns. May 2004;4(3):351-8. IF=2.293
Boon LM, Mulliken JB, Enjolras O, Vikkula M. Glomuvenous malformation (glomangioma) and venous malformation: distinct clinicopathologic and genetic entities. Arch Dermatol. Aug 2004;140(8):971-6. IF=3.187
Boon LM, Mulliken JB, Vikkula M. RASA1: variable phenotype with capillary and arteriovenous malformations. Curr Opin Genet Dev. Jun 2005;15(3):265-9. IF=9.361
Ghassibe M, Bayet B, Revencu N, Verellen-Dumoulin C, Gillerot Y, Vanwijck R, Vikkula M. Interferon regulatory factor-6: a gene predisposing to isolated cleft lip with or without cleft palate in the Belgian population. Eur J Hum Genet. Nov 2005;13(11):1239-42. IF=3.251
Gutierrez-Roelens I, Sluysmans T, Jorissen M, Amyere M, Vikkula M. Localization of candidate regions for a novel gene for Kartagener syndrome. Eur J Hum Genet. Jul 2006;14(7):809-15. IF=3.697
Ghalamkarpour A, Morlot S, Raas-Rothschild A, Utkus A, Mulliken JB, Boon LM, Vikkula M. Hereditary lymphedema type I associated with VEGFR3 mutation: the first de novo case and atypical presentations. Clin Genet. Oct 2006;70(4):330-5. IF=3.140
Ghassibe M, Bernier V, Boon LM, Vikkula M. A novel mutation in the MSX2 homeobox gene of a family with foramina parietalia permagna, headache and vascular anomaly. Eur J Pediatr. Oct 2006;165(10):734-735. IF=1.137
Gutierrez-Roelens I, De Roy L, Ovaert C, Sluysmans T, Devriendt K, Brunner HG, Vikkula M. A novel CSX/NKX2-5 mutation causes autosomal-dominant AV block: are atrial fibrillation and syncopes part of the phenotype ? Eur J Hum Genet. Dec 2006;14(12):1313-1316. IF=3.697
Mallory SB, Enjolras O, Boon LM, Rogers E, Berk DR, Blei F, Baselga E, Ros AM, Vikkula M. Congenital plaque-type glomuvenous malformations presenting in childhood. Arch Dermatol. Jul 2006;142(7):892-6. IF=2.851
Rousseau E, Palm Th, Scaravilli F, Ruchoux MM, Figerella-Branger D, Salmon I, Ellison D, Lacroix C, Chapon F, Mikol J, Vikkula M, Godfraind C. Trisomy 19 ependymoma, a newly recognized genetico-histological association, including clear cell ependymoma. Mol Cancer 2007, 6(47). (two last authors contributed equally) IF=0.756
Limaye N, Revencu N, Van Regemorter N, Garzon M, Bonduelle M, Chung W, Daras MD, Fahey MC, Garrett C, Gillerot Y, Gillessen-Kaesbach G, Gimenez-Arnau A, Guzzetta F, Battaglia D, Heimdal K, Lissens W, Taub E, Van Maldergem L, Van Paesschen W, Wieczorek, D, Wood NW, Boon L, Vikkula M. Novel human pathological mutations. Gene symbol: KRIT1. Disease: cerebral cavernous malformation. Hum Genet 2007, 122(5):552. IF=3.844
Revencu N, Boon LM, Mulliken JB, Enjolras O, Cordisco MR, Burrows PE, Clapuyt, Hammer F, Dubois J, Baselga E, Brancati F, Carder R, Ceballos Quintal JM, Dallapiccola B, Fischer G, Frieden IJ, Garzon M, Harper J, Johnson-Patel J, Labrèze C, Martorell L, Paltiel H, Pohl A, Prendiville J, Quere I, Siegel DH, Valente EM, van Hagen A, van Hest L,

Vaux KK, Vicente A, Weibel L, Chitayat D, Vikkula M. Parkes Weber syndrome, vein of Galen aneurysmal malformation, and other fast-flow vascular anomalies and specific neural tumors associated with RASA1 mutations. Hum Mutat 2008, 29(7):959-65. IF=7.033
Coutelier M, Andries S, Ghariani S, Dan B, Duyckaerts C, van Rijckevorsel K, Raftopoulos C, Deconinck N, Sonderegger P, Scaravilli F, Vikkula M*, Godfraind C*. Neuroserpin mutation causes electrical status epilepticus of slow-wave sleep. Neurology 2008, 71(1) :64-6. IF=7.043
Persu A, Hamoir M, Grégoire V, Garin P, Duvivier E, Reychler H, Chantrain G, Mortier G, Mourad M, Maiter D, Vikkula M. High prevalence of SDHB mutations in head and neck paraganglioma in Belgium. J Hyperten 2008, 26(7) :1395-1401. IF=5.132
Persu A, Amyere M, Gutierrez-Roelens I, Sempoux C, Lecouvet FE, Van Beers BE, Horsmans Y, De Plaen J-F, Hamoir M, Vikkula M. Rare presentation of familial paraganglioma without evidence of mutation in the SDH, RET and VHL genes : towards further genetic heterogenity. J Hypertension 2009, 27:76-82. IF=4.988
Limaye N*, Wouters V*, Uebelhoer M, Tuominen M, Wirkkala R, Mulliken JB, Eklund L, Boon LM, Vikkula M. Somatic mutations in angiopoïetin-receptor gene TEK cause solitary and multiple sporadic venous malformations. Nat Genet, 2009, 41(1):118-124. (*Underlined authors contributed equally to this work). IF=34.284.
Ghalamkarpour A, Holnthoner W, Saharinen P, Boon LM, Mulliken JB, Alitalo K, Vikkula M. Recessive primary congenital lymphoedema caused by a VEGFR3 mutation. J Med Genet 2009, 46(6) : 399-404. IF=5.751
Ghalamkarpour A, Debauche Ch, Haan E, Van Regemorter N, Sznajer Y, Thomas D, Revencu N, Gillerot Y, Boon LM, and Vikkula M. Sporadic in utero generalized edema associated with mutations in the lymphangiogenic genes VEGFR3 and FOXC2 (short report). J Pediatrics 2009, 155(1) :90-93. IF=4.092
Palm T, Figarella-Branger D, Chapon F, Lacroix C, Gray F, Scaravilli F, Vandenbroecke C, Ellison D, Salmon I, Vikkula M*, Godfraind C*. Expression profiling of ependymomas unravels localisation and tumor grade specific tumorigenesis. Cancer 2009, 115(17):3955-3968.*These authors contributed equally to this work. IF=5.418
Godfraind C and Vikkula M. Nervous system: Trisomy 19 ependymoma. Atlas Genet Cytogenet Oncol Haematol 2009, 13(1):90-92.
Wouters V, Limaye N, Uebelhoer M, Irrthum A, Boon LM, Mulliken JB, Enjolras O, Baselga E, Berg J, Dompmartin A, Ivarsson SA, Kangesu L, Lacassie Y, Murphy J, Teebi AS, Pennington A, Rieu P, Vikkula M. Hereditary cutaneomucosal venous malformations are caused by TIE2 mutations with widely variable hyper-phosphorylating effects. Eur J Hum Genet 2010, Apr; 18(4):414-420. IF=4.380
Desmyter L, Ghassibe M, Revencu R, Boute O, Lees M, François G, Verellen-Dumoulin C, Sznajer Y, Moncla A, Benateau H, Claes K, Devriendt K, Mathieu M, Van Maldergem L, Addor MC, Drouin-Garraud V, Mortier G, Bouma M, Dieux-Coeslier A, Genevieve D, Goldenberg A, Gozu A, Makrythanasis P, McEntagart M, Sanchez A, Vilain C, Vermeer S, Connell F, Verheij J, Manouvrier S, Pierquin G, Odent S, Holder-Espinasse M, Vincent-Delorme C, Gillerot Y, Vanwijck R, Bayet B and Vikkula M. IRF6 screening of syndromic and a priori non-syndromic cleft lip and palate patients: identification of a new type of minor VWS sign. Mol Syndromol 2010, 1:67-74.
Ghassibe-Sabbagh M, Desmyter L, Langenberg T, Hermans K, Boute O, Bayet B, Pellerin Ph, Claes F, Backx L, Mansilla MA, Imoehl S, Nowak S, Ludwig KU, Baluardo C, Ferrian M, Mossey PA, Noethen M, Dewerchin M, François G, Revencu N, Vanwijck R, Hecht J, Mangold E, Murray J, Rubini M, Vermeesch JR, Antoine-Poirel H, Carmeliet P, Vikkula M. FAF1, a gene that is disrupted in cleft palate and has conserved function in Zebrafish. Am J Hum Genet 2011, 88(2) :150-61. IF=10.603
Amyere M, Vogt Th, Hoo J, Brandrup F, Boon LM, Vikkula M. KITLG mutations cause familial progressive hyper- and hypo-pigmentation. J Invest Dermatol, 2011, 131(6) :1234-9. IF=6.314
Persu A and Vikkula M. A génome-wide association study-derived candidate gene seeks réplication : STK39. J Hypertens 2011, 29(3) :484-91. IF=4.021
Persu A, Lannoy N, Maiter D, Mendola A, Montigny P, Oriot P, Vinck W, Garin P, Hamoir M, Vikkula M. Prevalence and spectrum of SDHx mutations in pheochromocytoma and paraganglioma in patients from Belgium: an update. Horm Metab Res 2012, 44(5):349-53. IF=2.188
Brouillard P, Boon LM, Revencu N, Berg J, Dompmartin A, Dubois J, Garzon M, Holden S, Kangesu L, Labrèze C, Lynch SA, McKeown, Meskauskas R, Quere I, Syed S, Vabres P, Wassef M, Mulliken JB, Vikkula M and the GVM-study group. Genotypes and phenotypes of 162 families with a glomulin mutation. Mol Syndromol 2013, 4(4) :157-164.
Revencu N, Boon LM, Dompmartin A, Rieu P, Busch WL, Dubois J, Forzano F, van Hagen JM, Halbach S, Kuechler A, Lachmeijer AM, Lähde J, Russell L, Simola KO, Mulliken JB, Vikkula M. Germline mutations in RASA1 are not found in patients with Klippel-Trenaunay syndrome or capillary malformation with limb overgrowth. Mol Syndromol 2013, 4(4) :173-178.
Soblet J, Limaye N, Uebelhoer M, Boon LM, Vikkula M. Variable somatic TIE2 mutations in half of sporadic venous malformations. Mol Syndromol 2013, 4(4) :179-183.
Mendola A*, Schlögel M*, Ghalamkarpour A, Irrthum A, Nguyen HL, Fastré E, Yilmaz N, Duchateau T, Townshend S, Quichet A, Bygum A, Maat-Kievit A, van der Vleuten C, Fagerberg C, Jensen P, Coubes C, Mignot C, Baselga E, Inzana F, Repetto G, Garcia Ortiz JE, Lehtonen L, Wuyts W, Tiller GE, Wehner L-E, El-Khateeb MS, Legius E, Blei F, Quere I, Perdu J, Sarda P, Mulliken JB, Boon LM, Brouillard P, Vikkula M. Mutations in VEGFR3 signaling pathway explain 36% of primary lymphedema. Mol Syndromol 2013, 4 :257-266. (*Underlined authors contributed equally to this work).
Amyere M*, Aerts V*, Brouillard P*, McIntyre B, Duhoux F, Wassef M, Enjolras O, Mulliken JB, Antoine-Poirel H, Devuyst O, Boon LM, Vikkula M. Somatic uniparental isodisomy explains multifocality of glomuvenous malformations. Am J Hum Genet 2013, 92(2) :188-96. (*Underlined authors contributed equally to this work). IF=10.987
Uebelhoer M, Nätynki M, Kangas J, Mendola A, Nguyen H-L, Soblet J, Godfraind C, Boon LM, Eklund L, Limaye N, Vikkula M. Venous malformation-causative TIE2 mutations mediate an AKT-dependent decrease in PDGFB. Hum Mol Genet 2013, 22(17) :3438-48. IF=6.677
Revencu N, Boon LM, Mendola A, Dubois J, Clapuyt P, Hammer F, Amor DJ, Irvine AD, Baselga E, Dompmartin A, Syed S, Martin Santiago A, Ades L, Collins F, Smith J, Sandaradura S, Barrio VR, Burrows P, Blei F, Cozzolino M, Brunetti-Pierri N, Vicente A, Abramowicz, Désir J, Vilain C, Chung WK, Wilson A, Gardiner CA, Dwight Y, Lord DJE, Fishman L, Cytrynbaum C, Chamlin S, Ghali F, Gilaberte Y, Joss S, del C. Boente M, Léauté-Labrèze C, Delrue M-A, Bayliss S, Martorell L, Gonzalez M-A, Mazereeuw-Hautier J, O'Donnell B, Bessis D, Pyeritz RE, Salhi A, Tan OT, Mulliken JB, Vikkula M. RASA1mutations and associateed phenotypes in 68 families with capillary malformation-arteriovenous malformation. Hum Mutat 2013, 34(12) :1632-41. IF=5.122
Amyere M, Dompmartin A, Wouters V, Enjolras O, Kaitila I, Docquier P-L, Godfraind C, Mulliken JB, Boon LM, Vikkula M. Common somatic alterations identified in Maffucci syndrome by molecular keryotyping. Mol Syndromol 2014, 5(6) :259-67. IF=3.17
Boscolo E, Limaye N, Huang L, Kang KT, Soblet J, Uebelhoer M, Mendola A, Natynki M, Seront E, Dupont S, Hammer J, Legrand C, Brugnara C, Eklund L, Vikkula M*, Bischoff J*, Boon LM*. Rapamycin improves TIE2-mutated venous malformation in murine model and human subjects. J Clin Invest 2015 Sep 1;125(9):3491-504. (*Underlined authors contributed equally to this work). IF=12.575
Schlögel M, Mendola A, Fastré E, Vasudevan P, Devriendt K, de Ravel TJL, Van Esch H, Casteel I, Arroyo Carrera IA, Cristofoli F, Fieggen K, Jones K, Lipson M, Singer A, Soller M, Villanueva MM, Revencu N, Boon LM, Brouillard P, Vikkula M. No evidence of locus heterogeneity in familial microcephaly with or without chorioretinopathy, lymphedema, or mental retardation syndrome. Orphanet J Rare Dis (OJRD) 2015, 10(1): 52. IF2014=3.358
Persu A, Evenepoel L, Jin Y, Mendola A, Staessen JA, Vikkula M. 6C.05: STK39 and WNK1 are potential hypertension susceptibility genes in the BELHYPGEN cohort. J Hypertens 2015 Jun;33 Suppl 1:e80. doi: 10.1097/01.hjh.0000467569.10961.69.
Ballieux F, Boon LM, Vikkula M. Blue Bleb Rubber Nevus Syndrome. Handb Clin Neurol 2015;132:223-30. IF=2.22
Boon LM, Hammer J, Seront E, Dupont S, Hammer F, Clapuyt P, Vikkula M. Rapamycin as novel treatment for refractory-to-standard-care slow-flow vascular malformations. Plast Reconstr Surg 2015 Oct;136(4 Suppl):38. IF=3.09
Limaye N, Kangas J, Mendola A, Godfraind C, Schlögel MJ, Helaers R, Eklund L, Boon LM, Vikkula M. Somatic activating PIK3CA mutations cause venous malformation. Am J Hum Genet 2015, Dec 3;97(6):914-21. IF=10.48
Seront E, Limaye N, Boon LM, Vikkula M. La rapamycine ouvre l'ère des thérapies ciblées dans les malformations

veineuses. Médicine/Sciences 2016, 32(6-7) :574-8.
Soblet J, Kangas J, Nätynki M, Mendola A, Helaers R, Uebelhoer M, Kaakinen M, Cordisco M, Dompmartin A, Enjolras O, Holden S, Irvine AD, Kangesu L, Léauté-Labrèze Ch, Lanoel A, Lokmic Z, Maas S, McAleer MA, Penington A, Rieu P, Syed S, van der Vleuten C, Watson R, Fishman SJ, Mulliken JB, Eklund L, Limaye N, Boon LM, Vikkula M. Blue Rubber Bleb Nevus (BRBN) Syndrome is Caused by Somatic TEK (TIE2) Mutations. J Invest Dermatol 2017, 137(1) :207-16.
Poirel HA and Vikkula M. Age-related heterogeneity of Burkitt lymphoma : response to Mbulaiteye and Anderson. Br J Haematol, 2016, Sep 9. doi: 10.1111/bjh.14309.
Helaers R, Brouillard P, Soblet J, Amyere M, Limaye N, Vikkula M. Highlander: variant filtering made easy. Hum Mutat, resubmitted.
Amyere M*, Revencu N*, Helaers R, Pairet E, Baselga E, Cordisco M, Chung W, Dubois J, Lacour J-Ph, Martorell L, Mazereeuw-Hautier J, Pyeritz RE, Amor DJ, Bisdorff A, Blei F, Bombei H, Dompmartin A, Brooks D, Dupont J, González-Enseñat MA, Frieden I, Gérard M, Kvarnung M, Kwan Hanson-Kahn A, Hudgins L, Léauté-Labrèze Ch, McCuaig C, Metry D, Parent Ph, Paul C, Petit F, Phan A, Quere I, Salhi A, Turner A, Vabres P, Vicente A, Wargon O, Watanabe S, Weibel L, Wilson A, Willing M, Mulliken JB, Boon LM, Vikkula M. Germline loss-of-function mutations in EPHB4 cause a second form of capillary malformation–arteriovenous malformation (CM-AVM2) deregulating RAS-MAPK signaling. Circulation 2017, 136(11) :1037-48. (*Underlined authors contributed equally to this work). IF=18.88
Nguyen H-L, Boon LM, Vikkula M. Vascular anomalies caused by abnormal signaling within endothelial cells : targets for novel therapies. Semin Intervent Radiol 2017, 34(3) :233-8. IF=2.41
Brouillard P, Dupont L, Helaers R, Coulie R, Tiller GE, Peeden J, Colige A, Vikkula M. Loss of ADAMTS3 activity causes Hennekam lymphangiectasia-lymphedema syndrome 3. Hum Mol Genet 2017, 26(21) :4095-104. IF=4.90
Basha M, Demeer B, Revencu N, Helaers R, Theys S, Bou Saba S, Boute O, Devauchelle B, Francois G, Bayet B, Vikkula M. Whole exome sequencing identifies mutations in 10% of patients with familial non-syndromic cleft lip and/or palate in genes mutated in well-known syndromes. J Med Genet 2018, 55(7) :449-58. IF=5.451
Fastré E, Lanteigne L-E, Helaers R, Giacalone G, Revencu N, Dionyssiou D, Demiri E, Brouillard P, Vikkula M. Splice-site mutations in VEGFC cause loss-of-function and Nonne-Milroy-like primary lymphedema. Clin Genet 2018, 94(1) :179-81. IF=3.51
Quéré I, Nagot N, Vikkula M. Incidence of cellulitis among children with primary lymphedema. New Engl J Med 2018, 378(21):2047-8. IF2018=70.670
Demeer B, Revencu N, Helaers R, Devauchelle B, François G, Bayet B, Vikkula M. Unmasking familial CPX by WES and identification of novel clinical signs. Am J Med Genet 2018, 176(12) :2661-7.
Seront E, Van Damme A, Boon LM, Vikkula M. Rapamycine and treatment of venous malformations (review). Curr Op Hematol 2019, May;26(3):185-192.
Demeer B, Revencu N, Helaers R, Gbaguidi C, Dakpe S, François G, Devauchelle B, Bayet B, Vikkula M. Likely pathogenic variants in one third of non-syndromic discontinuous cleft lip and palate patients. Genes (Basel) 2019 Oct 22;10(10), pii:E833.
Revencu N, Fastré E, Ravoet M, Helaers R, Brouillard P, Bisdorff-Breson A, Chung CWT, Gerard M, Dvoravoka V, Irvine AD, Boon, LM, Vikkula M. RASA1 mosaic mutations in patients with capillary malformation – arteriovenous malformation. J Med Genet 2019, 57(1):48-52.
d'Avila-Levy CM, Bearzatto B, Ambroise J, Helaers R, Butenko A, Yurchenko V, Morelli KA, Santos HLC, Brouillard P, Grellier P, Gala J-L, Vikkula M. First draft genome of the trypanosomatid Herpetomonas muscarum ingenoplastis through MinION Oxford Nanopore technology and Illumina sequencing. Trop Med and Infect Dis 2020, 5(1):E25.
Leppänen VM*, Brouillard P*, Korhonen EA, Sipilä T, Jha S, Revencu N, Labarque V, Fastré E, Schlögel M, Ravoet M, Singer A, Luzzatto C, Angelone D, Crichiutti G, D'Elia A, Kuurne J, Elamaa H, Koh GY, Saharinen P, Vikkula M* and Alitalo K* (*Underlined authors contributed equally to this work). Characterization of ANGPT2 mutations associated with primary lymphedema. Sci Trans Med 2020.
Dekeuleneer V, Seront E, Van Damme A, Boon LM, Vikkula M. Theranostic Advances in Vascular Malformations. J Invest Dermatol. 2020 Apr;140(4):756-763.
Nguyen HL, Boon LM, Vikkula M. Genetics of vascular anomalies. Semin Pediatr Surg. 2020 Oct;29(5):150967.
Van Damme A, Seront E, Dekeuleneer V, Boon LM, Vikkula M. New and Emerging Targeted Therapies for Vascular Malformations. Am J Clin Dermatol. 2020 Oct;21(5):657-668.
Revencu N, Fastre E, Ravoet M, Helaers R, Brouillard P, Bisdorff-Bresson A, Chung CWT, Gerard M, Dvorakova V, Irvine AD, Boon LM, Vikkula M. RASA1 mosaic mutations in patients with capillary malformation-arteriovenous malformation. J Med Genet. 2020 Jan;57(1):48-52.
Brouillard P*, Schlögel MJ*, Homayun Sepehr N, Helaers R, Queisser A, Fastré E, Boutry S, Schmitz S, Clapuyt P, Hammer F, Dompmartin A, Weitz-Tuoretmaa A, Laranne J, Pasquesoone L, Vilain C, Boon LM, Vikkula M. Non-hotspot PIK3CA mutations are more frequent in CLOVES than in common or combined lymphatic malformations. Orphanet J Rare Dis. 2021 Jun 10;16(1):267. (*Underlined authors contributed equally to this work).
Queisser A, Seront E, Boon LM, Vikkula M. Genetic Basis and Therapies for Vascular Anomalies. Circ Res. 2021 Jun 25;129(1):155-173.
Brouillard P, Witte MH, Erickson RP, Damstra RJ, Becker C, Quéré I, Vikkula M. Primary lymphoedema. Nat Rev Dis Primers. 2021 Oct 21;7(1):77.
Smeland MF, Brouillard P, Prescott T, Boon LM, Hvingel B, Nordbakken CV, Nystad M, Holla ØL, Vikkula M. Biallelic ANGPT2 loss-of-function causes severe early-onset non-immune hydrops fetalis. J Med Genet. 2021 Dec 7:jmedgenet-2021-108179
Greene AK, Brouillard P, Sudduth CL, Smits PJ, Konczyk DJ, Vikkula M. EPHB4 mutation causes adult and adolescent-onset primary lymphedema. Am J Med Genet A. 2021 Dec;185(12):3810-3813.
Coulie J, Boon L, Vikkula M. Molecular Pathways and Possible Therapies for Head and Neck Vascular Anomalies. J Oral Pathol Med. 2022 May 24.
L. M. Boon, V. Dekeuleneer, J. Coulie, L. Marot, A.-C. Bataille, F. Hammer, Philippe Clapuyt, Anne Jeanjean, Anne Dompmartin & Vikkula M. , Case report study of thalidomide therapy in 18 patients with severe arteriovenous malformations, Nature Cardiovascular Research 2022 ,Vol 1, June 2022, 562–567.
Diociaiuti A, Baselga E, Boon LM, Dompmartin A, Dvorakova V, El Hachem M, Gasparella P, Haxhija E, Ghaffarpour N, Kyrklund K, Irvine AD, Kapp FG, Rößler J, Salminen P, van den Bosch C, van der Vleuten C, Kool LS, Vikkula M. The VASCERN-VASCA working group diagnostic and management pathways for severe and/or rare infantile hemangiomas. Eur J Med Genet. 2022 Jun;65(6):104517.
Cordisco M.R., El-Feghaly J., Prezzano J.C., Lanöel A., Torres N., Persico, S., Requejo F.,Sierre S., Fiandrino M.J., Luna L., Maccario M.F.,Brouillard, P. Vikkula M. Capillary Malformation-Arteriovenous Malformation Type 2, A Report of 6 Cases and Main Differential Diagnosis. Journal of Vascular Anomalies 3(4):p e053, December 2022.
Smeland MF, Brouillard P, Prescott T, Boon LM, Hvingel B, Nordbakken CV, Nystad M, Holla ØL, Vikkula M. Biallelic ANGPT2 loss-of-function causes severe early-onset non-immune hydrops fetalis. J Med Genet. 2023 Jan;60(1):57-64.
Nguyen HL, Boon LM, Vikkula M. Trametinib as a promising therapeutic option in alleviating vascular defects in an endothelial KRAS-induced mouse model. Hum Mol Genet. 2023 Jan 6;32(2):276-289.
Boutry S, Helaers R, Lenaerts T, Vikkula M. A new statistical framework for rare-variant association testing. PLOS Computational Biology, 2023 19(9): e1011488.
Boutry S, Helaers R, Lenaerts T, Vikkula M. Rare variant association on unrelated individuals in case–control studies using aggregation tests: existing methods and current limitations. Briefings in Bioinformatics, Volume 24, Issue 6, November 2023.
De Bortoli M, Ivars M, Revencu N, Nassogne MC, Lavarino C, Paco S, Lammens M, Renders A, Dumitriu D, Helaers R, Boon LM, Baselga E, Vikkula M. Epilepsy with faint capillary malformation or reticulated telangiectasia associated with mosaic AKT3 pathogenic variants. Am J Med Genet A. 2024 Feb 6:e63551.
De Bortoli M, Queisser A, Pham VC, Dompmartin A, Helaers R, Boutry S, Claus C, De Roo AK, Hammer F, Brouillard P, Abdelilah-Seyfried S, Boon LM, Vikkula M. Somatic loss-of-function PIK3R1 and activating non-hotspot PIK3CA mutations associated with Capillary Malformation with Dilated Veins (CMDV). J Invest Dermatol. 2024 Feb 29:S0022-202X(24)00164-7.
Alpaslan M, Fastré E, Mestre S, van Haeringen A, Repetto GM, Keymolen K, Boon LM, Belva F, Giacalone G, Revencu

N, Sznajer Y, Riches K, Keeley V, Mansour S, Gordon K, Martin-Almedina S, Dobbins S, Ostergaard P, Quere I, Brouillard P, Vikkula M. Pathogenic variants in HGF give rise to childhood-to-late onset primary lymphoedema by loss of function. Hum Mol Genet. 2024 Jul 6;33(14):1250-1261.
Revencu N, Eijkelenboom A, Bracquemart C, Alhopuro P, Armstrong J, Baselga E, Cesario C, Dentici ML, Eyries M, Frisk S, Karstensen HG, Gene-Olaciregui N, Kivirikko S, Lavarino C, Mero IL, Michiels R, Pisaneschi E, Schönewolf-Greulich B, Wieland I, Zenker M, Vikkula M. Assessment of gene-disease associations and recommendations for genetic testing for somatic variants in vascular anomalies by VASCERN-VASCA. Orphanet J Rare Dis. 2024 May 22;19(1):213.
Brouillard P, Murtomäki A, Leppänen VM, Hyytiäinen M, Mestre S, Potier L, Boon LM, Revencu N, Greene AK, Anisimov A, Salo MH, Hinttala R, Eklund L, Quéré I, Alitalo K, Vikkula M. Loss-of-function mutations of the TIE1 receptor tyrosine kinase cause late-onset primary lymphedema. J Clin Invest. J Clin Invest. 2024;134(14):e173586.
Revencu N, Eijkelenboom A, Bracquemart C, Alhopuro P, Armstrong J, Baselga E, Cesario C, Dentici ML, Eyries M, Frisk S, Karstensen HG, Gene-Olaciregui N, Kivirikko S, Lavarino C, Mero IL, Michiels R, Pisaneschi E, Schönewolf-Greulich B, Wieland I, Zenker M, Vikkula M. Correction to: Assessment of gene-disease associations and recommendations for genetic testing for somatic variants in vascular anomalies by VASCERN-VASCA. Orphanet J Rare Dis. 2024 Oct 7;19(1):369. doi: 10.1186/s13023-024-03296-6. Erratum for: Orphanet J Rare Dis. 2024 May 22;19(1):213.
Seront, E., Queisser, A., Boon, L. M., & Vikkula, M. (2024). Molecular landscape and classification of vascular anomalies. Hematology, 2024(1), 700-708.
Ranji P, Pairet E, Helaers R, Bayet B, Gerdom A, Gil-da-Silva-Lopes VL, Revencu N, Vikkula M. Four putative pathogenic ARHGAP29 variants in patients with non-syndromic orofacial clefts (NsOFC). Eur J Hum Genet. 2025 Jan;33(1):38-43.
Baselga, E., Andersen, R., Barea, M., Bejarano Serrano, M., Berger, S., Bisdorff-Bresson, A., Boccara, O., Bom-Sucesso, M., Boon, L. M., Borgards, P., Diociaiuti, A., Dompmartin, A., Dvorakova, V., El Hachem, M., Frisk, S., Gasparella, P., Ghaffarpour, N., Haxhija, E., Hjuler, T., … Vikkula, M. (2025). The VASCERN-VASCA Working Group Diagnostic and Management Pathways for Capillary Malformations. Journal of Vascular Anomalies, 6(1).
Ranji P, Pairet E, Helaers R, Brouillard P, Bayet B, Gerdom A, Revencu N, Vikkula M. Likely Pathogenic/Pathogenic Variants in the Spliceosome Complex Genes SNRNP200, SF3B1, SF3B2, and SF3B4 Implicated in Nonsyndromic Orofacial Cleft. Hum Mutat. 2025 Dec 14;2025:2991452.
Gasparella P, Haxhija EQ, Andersen R, Barea M, Baselga E, Serrano MB, Berger S, Bisdorff AA, Boccara O, Borgards P, Bom-Sucesso M, Boon LM, Cimpean AM, Diociaiuti A, Dvorakova V, Hachem ME, Frisk S, Ghaffarpour N, Holm A, Irvine AD, Kaltoft M, Kapp FG, Koskova O, Kyrklund K, Madureira M, Palionis D, Przewratil P, Schönewolf-Greulich B, Stanciulescu MC, Štěrba J, Tolonen J, Vaisnyte B, van der Vleuten C, Wyrzykowski D, Kool LS, Vikkula M. The VASCERN-VASCA diagnostic and management pathways for kaposiform hemangioendothelioma. Eur J Pediatr. 2025 Dec 13;185(1):14.

B. Articles (co-author)

Palotie A, Vaisanen P, Ott J, Ryhänen L, Elima K, Vikkula M, Cheah K, Vuorio E, Peltonen L. Predisposition to familial osteoarthrosis linked to type II collagen gene. Lancet. Apr 29 1989;1(8644):924-7. IF=14.435
Boon LM, Mulliken JB, Vikkula M, Watkins H, Seidman J, Olsen BR, Warman ML. Assignment of a locus for dominantly inherited venous malformations to chromosome 9p. Hum Mol Genet. Sep 1994;3(9):1583-7. IF=4.528
Gallione CJ, Pasyk KA, Boon LM, Lennon F, Johnson DW, Helmbold EA, Markel DS, Vikkula M, Mulliken JB, Warman ML, Pericak-Vance MA, Marchuk DA. A gene for familial venous malformations maps to chromosome 9p in a second large kindred. J Med Genet. Mar 1995;32(3):197-9. IF=2.701
Ritvaniemi P, Korkko J, Bonaventure J, Vikkula M, Hyland J, Paassilta P, Kaitila I, Kääriäinen H, Sokolov BP, Hakala M, Mannismäki P, Meerson EM, Klemola T, Williams C, Peltonen L, Kivirikko KI, Prockop DJ, Ala-Kokko L. Identification of COL2A1 gene mutations in patients with chondrodysplasias and familial osteoarthritis. Arthritis Rheum. Jul 1995;38(7):999-1004. IF=7.232
Gong Y, Vikkula M, Boon LM, Liu J, Beighton P, Ramesar R, Peltonen L, Somer H, Hirose T, Dallapiccola B, de Paepe A, Swoboda W, Zabel B, Superti-Furga A, Steinmann B, Brunner H, Jans A, Boles RG, Adkins W, van den Boogaard M-J, Olsen BR, Warman ML. Osteoporosis-pseudoglioma syndrome, a disorder affecting skeletal strength and vision, is assigned to chromosome region 11q12-13. Am J Hum Genet. Jul 1996;59(1):146-51. IF=9.366
Veiga-da-Cunha M, Gerin I, Chen YT, de Barsy Th, de Lonlay P, Dionisi-Vici C, Fenske CD, Lee PJ, Leonard JV, Maire I, McConkie-Rosell A, Schweitzer S, Vikkula M, Van Schaftingen E. A gene on chromosome 11q23 coding for a putative glucose- 6-phosphate translocase is mutated in glycogen-storage disease types Ib and Ic. Am J Hum Genet. Oct 1998;63(4):976-83. IF=10.869
Korpelainen EI, Karkkainen M, Gunji Y, Vikkula M, Alitalo K. Endothelial receptor tyrosine kinases activate the STAT signaling pathway: mutant Tie-2 causing venous malformations signals a distinct STAT activation response. Oncogene. Jan 7 1999;18(1):1-8. IF=6.517
Annunen S, Korkko J, Czarny M, Warman ML, Brunner HG, Kääriäinen H, Mulliken JB, Tranebjaerg L, Brooks DG, Cox GF, Cruysberg JR, Curtis MA, Davenport SLH, Friedrich CA, Kaitila I, Krawczynski MR, Latos-Bielenska A, Mukai S, Olsen BR, Shinno N, Somer M, Vikkula M, Zlotogora J, Prockop DJ, Ala-Kokko L. Splicing mutations of 54-bp exons in the COL11A1 gene cause Marshall syndrome, but other mutations cause overlapping Marshall/Stickler phenotypes. Am J Hum Genet. Oct 1999;65(4):974-83. IF=10.426
Veiga-da-Cunha M, Gerin I, Chen YT, Lee PJ, Leonard JV, Maire I, Wendel U, Vikkula M, Van Schaftingen E. The putative glucose 6-phosphate translocase gene is mutated in essentially all cases of glycogen storage disease type I non-a. Eur J Hum Genet. Sep 1999;7(6):717-23. IF=2.554
Baurain JF, Colau D, van Baren N, Landry C, Martelange V, Vikkula M, Boon T, Coulie PG. High frequency of autologous anti-melanoma CTL directed against an antigen generated by a point mutation in a new helicase gene. J Immunol. Jun 1 2000;164(11):6057-66. IF=6.834
Gong Y, Slee RB, Fukai N, Rawadi G, Roman-Roman S, Reginato A, Wang H, Cundy T, Glorieux F, Lev D, Zacharin M, Oexle K, Marcelino J, Suwairi W, Heeger S, Sabatakos G, Apte S, Adkins WN, Allgrove J, Arslan-Kirchner M, Batch JA, Beighton P, Black GCM, Boles RG, Boon LM, Borrone C, Brunner H, Carle GF, Dallapiccola B, De Paepe A, Floege B, Lees Halfhide M, Hall B, Hennekam RC, Hirose T, Jans A, Jüppner H, Kim C, Keppler-Noreuil K, LaCombe D, Lambert M, Lemyre E, Letteboer T, Peltonen L, Ramesar RS, Romanengo M, Somer H, Steichen-Gersdorf E, Steinmann B, Sullivan B, Superti-Furga A, Swoboda W, van den Boogaard M-J, Van Hul W, Vikkula M, Votruba M, Zabel B, Garcia T, Baron R, Olsen BR, Warman ML. LDL receptor-related protein 5 (LRP5) affects bone accrual and eye development. Cell. Nov 16 2001;107(4):513-23. IF=29.219
Rzem R, Veiga-da-Cunha M, Noel G, Goffette S, Nassogne M-C, Tabarki B, Schöller C, Marquardt T, Vikkula M, Van Schaftingen E. A gene encoding a putative FAD-dependent L-2-hydroxyglutarate dehydrogenase is mutated in L-2-hydroxyglutaric aciduria. Proc Natl Acad Sci USA. Nov 30 2004;101(48):16849-54. IF=10.452
Zanardo L, Stolz W, Schmitz G, Kaminski W, Vikkula M, Landthaler M, Vogt T. Progressive hyperpigmentation and generalized lentiginosis without associated systemic symptoms: a rare hereditary pigmentation disorder in south-east Germany. Acta Derm Venereol. 2004;84(1):57-60. IF=1.585
Daniel-Spiegel E, Ghalamkarpour A, Spiegel R, Weiner E, Vikkula M, Shalev E, Shalev SA. Hydrops fetalis: an unusual prenatal presentation of hereditary congenital lymphedema. Prenat Diagn. Nov 2005;25(11):1015-18. IF=1.640
Lorenz-Depiereux B, Bastepe M, Benet-Pages A, Amyere M, Wagenstaller J, Muller-Barth U, Badenhoop K, Kaiser SM, Rittmaster RS, Shlossberg AH, Olivares JL, Loris C, Ramos FJ, Glorieux F, Vikkula M, Jüppner H, Ström TM. DMP1 mutations in autosomal recessive hypophosphatemia implicate a bone matrix protein in the regulation of phosphate homeostasis. Nat Genet. Nov 2006;38(11):1248-1250. IF=24.176
Spiegel R, Ghalamkarpour A, Daniel-Spiegel E, Vikkula M, Shalev SA. Wide clinical spectrum in a family with hereditary lymphedema type I due to a novel missense mutation in VEGFR3. J Hum Genet. 2006;51(10):846-50. IF=2.205

Matsson H, Eason J, Bookwalter CS, Klar J, Gustavsson P, Sunnegårdh J, Enell H, Jonzon A, Vikkula M, Gutierrez I, Granados Riveron J, Pope M, Bu'Lock F, Cox J, Robinson TE, Song F, Brook JD, Marston S, Trybus KM, Dahl N. Alpha-cardiac actin mutations produce atrial septal defects. Hum Mol Genet 2008, 17:256-65. IF=7.249
Balikova I, Martens K, Melotte C, Amyere M, Van Vooren S, Moreau Y, Vetrie D, Fiegler H, Carter NP, Liehr T, Vikkula M, Matthijs G, Fryns J-P, Casteels I, Devriendt K, Vermeesch JR. Autosomal dominant microtia linked to five tandem copies of a copy number variable region at chromosome 4p16. Am J Hum Genet 2008, 82(1):181-187. IF=10.153
Dompmartin A, Acher A, Thibon P, Tourbach S, Hermans C, Deneys V, Pocock B, Lequerrec A, Labbé D, Barrellier M-T, Vanwijck R, Vikkula M, Boon LM. Association of localized intravascular coagulopathy with venous malformations. Arch Dermatol 2008, 144(7):873-7. IF=3.402
Couvineau A, Wouters V, Bertrand G, Rouyer C, Gérard B, Boon LM, Grandchamp B, Vikkula M, Silve C. PTHR1 mutations associated with Ollier disease result in receptor loss of function. Hum Mol Genet, 2008, 17(18) :2766-75. IF=7.249
Ben Rebeh I, Benzina Z, Dhouib H, Hadjamor I, Amyere M, Ayadi L, Turki K, Hammami B, Kmiha N, Kammoun H, Hakim B, Charfedine I, Vikkula M, Ghorbel A, Ayadi H, Masmoudi S. Identification of candidate regions foe a novel Usher syndrome type II locus. Molecular Vision 2008, 14 :1719-1726. IF=2.464
Jinnin M, Medici D, Park L, Limaye N, Liu Y, Boscolo E, Bischoff J, Vikkula M, Boye E, Olsen BR. Suppressed NFAT-dependent VEGFR1 expression and constitutive VEGFR2 signaling in infantile hemangioma. Nat Med, 2008, 14(11) :1236-46. IF=27.553
Ferreira de Lima RLL, Hoper SA, Ghassibe M, Cooper M, Rorick NK, Kondo S, Katz L, Marizita M, Compton J, Hehr-Regensburg U, Dixon MJ, Boute O, Bayet B, Revencu N, Verellen-Dumoulin C, Vikkula M, Richieri-Costa A, Moretti-Ferreira D, Murray JC, Schutte BC. Prevalence and non-random distribution of exonic mutations in Interferon Regulatory Factor 6 (IRF6) in 307 cases of Van der Woude syndrome and 37 cases of popliteal pterygium syndrome. Genet Med 2009, 11(4) :241-247. IF=3.922
Toll A, Parera E, Gimenez-Arnau AM, Pou A, Lloreta J, Limaye N, Vikkula M, Pujol RM. Cutaneous venous malformations in familial cerebral cavernomatosis caused by KRIT1 gene mutations. Dermatology 2009, 218(4):307-313. IF=2.741
Vanneste E, Voet Th, Le Caignec C, Ampe M, Konings P, Melotte C, Debrock S, Vikkula M, Schuit F, Fryns J-P, Verbeke G, D'Hooghe T, Moreau Y, Vermeesch J. Human cleavage stage embryos are chromosomally unstable. Nat Med, 2009, 15(5) :577-583. IF=27.136
Dompmartin A, Ballieux F, Thibon P, Lequerrec A, Hermans C, Clapuyt P, Barrellier M-T, Hammer F Labbé D, Vikkula M, Boon LM. Elevated D-dimer level in the differential diagnosis for venous malformations. Arch Dermatol, 2009, 145(11) :1239-44. IF=4.760
Wiame E, Tyteca D, Collard F, Amyere M, Noël G, Desmedt J, Nassogne M-C, Vikkula M, Courtoy P, Vincent M-F, Boltshauser E, Van Schaftingen E. Molecular identification of aspartate N-acetyltransferase and its mutation in hypoacetylaspartia. Biochem J 2009 Dec 14, 425(1) :127-136. IF=5.016
Alders M, Hogan BM, Salehi F, Al-Gazali L, Hennekam EA, Holmberg EE, Mannens MMAM, Mulder MF, Offerhaus GJ, Prescott TE, Schroor EJ, Verheij JB, Witte M, Zwijnenburg PJ, Vikkula M, Schulte-Merker S, Hennekam RC. Mutations in CCBE1 cause generalized lymph vessel dysplasia in humans. Nat Genet 2009 Dec, 41(12):1272-4. IF=34.284
Göhring I, Tagariello A, Endele S, Stolt CC, Ghassibé M, Fisher M, Thiel CT, Trautmann U, Vikkula M, Winterpacht A, Fitzpatrick DR, Rauch A. Disruption of ST5 is associated with mental-retardation and multiple congenital anomalies. J Med Genet 2010 Feb, 47(2):91-98. IF=7.037
Thiex R, Mulliken JB, Revencu N, Boon LM, Burrows PE, Cordisco MR, Dwight Y, Smith ER, Vikkula M, Orbach D. A novel association between RASA1 mutations and spinal arteriovenous anomalies. Am J Neuroradiol 2010 Apr, 31(4) :775-779. IF=3.464
Dompmartin A, Vikkula M, Boon LM. Venous malformation: update on etiopathogenesis, diagnosis & management. Phlebology 2010, 25:224-35. IF=2.698
Greenberger S, Reznik-Wolf H, Ghalamkarpour A, Marek D, Abu A, Trau H, Vikkula M, Pras E. Severe Congenital Lymphedema not caused by mutations in known lymphedema genes. Br J Dermatol 2010, 163(6) :1358-60. IF=4.353
Yao L, Schiavi F, Cascon A, Qin Y, Inglada LS, King EE, Toledo RA, Ercolino T, Rapizzi E, Ricketts CJ, Mori L, Giacchè M, Mendola A, Taschin E, Boaretto F, Loli P, Iacobone M, Rossi GP, Biondi B, Lima-Junior J, Kater CE, Bex M, Vikkula M, Grossman AB, Gruber S, Barontini M, Persu A, Castellano M, Toledo SPA, Maher ER, Mannelli M, Opocher G, Robledo M, Dahia PLM. Spectrum and prevalence of FP/TMEM127 gene mutations in pheochromocytomas and paragangliomas and insights into structure-function relantionship. JAMA 2010, 304(23) :2611-19. IF=30.011
Goujon E, Cordoro KM, Barat M, Rousseau T, Brouillard P, Vikkula M, Frieden IJ, Vabres P. Congenital plaque-type glomuvenous malformations associated with fetal pleural effusion and ascites. Ped Dermatol 2011, Sep-Oct;28(5):528-31. IF=1.072
Bowen ME, Boyden ED, Holm IA, Campos-Xavier B, Bonafé L, Superti-Furga A, Ikegawa S, Cormier-Daire V, Bovée JV, Pansuriya T, de Sousa SB, Savarirayan R, Andreucci E, Vikkula M, Garavelli L, Pottinger C, Ogino T, Sakai A, Regazzoni BM, Wuyts W, Sangiorgi L, Pedrini E, Zhu M, Kozakewich HP, Kasser JR, Seidman JG, Kurek KC, Warman ML. Loss-of-function mutations in PTPN11 cause metachondromatosis, but not Ollier disease or Maffucci syndrome. PLoS Genetics 2011, 7(4) :e1002050. IF=8.694
Carr CW, Zimmerman HH, Martin CL, Vikkula M, Byrd AC, Abdul-Rahman, OA. 5q14.3 neurocutaneous syndrome: a novel continguous gene syndrome caused by the simultaneous deletion of RASA1 and MEF2C. Am J Med Genet 2011, 155A(7):1640-5. IF=2.391
Pansuriya TC, van Eijk R, d'Adamo P, van Ruler MAJH, Kuijjer ML, Oosting J, Cleton-Jansen AM, van Oosterwijk JG, Verbeke S, Meijer D, van Wezel T, Nord KH, Sangiorgi L, Toker B, Liegl-Atzwanger B, San-Julian M, Sciot R, Limaye N, Kindblom L-G, Daugaard S, Godfraind C, Boon LM, Vikkula M, Kurek KC, Szuhai K, French PJ, Bovée JVMG. Somatic mosaic IDH1 or IDH2 mutations are associated with enchondroma and spindle cell hemangioma in Ollier disease and Maffucci syndrome. Nat Genet 2011, 43(12) :1256-61. IF-35.532
Dompmartin A, Blaizot X, Théron J, Hammer FM, Chene Y, Labbé D, Barrellier M-T, Gaillard C, Leroyer R, Chedru V, Ollivier C, Vikkula M, Boon LM. Radio-opaque ethylcellulose-ethanol is a safe and efficient sclerosing agent for venous malformations. Eur Radiol 2011, Dec ;21(12) :2647-56. IF=3.222
Duhoux F, Ameye G, Lambot V, Herens C, Lambert F, Raynaud S, Wlodarska I, Michaux L, Roche-Lestienne C, Labis E, Taviaux S, Chapiro E, Nguyen Khac F, Struski S, Dobbelstein S, Dastugue N, Lippert E, Cabrol C, Spelemen F, Van Roy N, De Weeer A, Rack K, Talmant P, Richebourg S, Mugneret F, Tigaud I, Mozziconacci MJ, Laibe S, Nadal N, Terré C, Libouton JM, Decottignies A, Vikkula M, A-Poirel H, on behalf of the Groupe Francophone de Cytogénétique Hématologique (GFCH) and of the Belgian Cytogenetic Group for Hematology and Oncology (BCG-HO). Refinement of 1p36 alterations not involving PRDM16 in myeloid and lymphoid malignancies. Plos One 2011, 6(10) :e26311. IF=4.092
Ostergaard P, Simpson MA, Mendola A, Vasudevan P, Connell FC, van Impel A, Moore AT, Loeys BL, Ghalamkarpour A, Onoufriadis A, Martinez-Corral I, Devery S, Leroy JG, van Laer L, Singer A, Bialer MG, McEntagart M, Quarrell O, Brice G, Trembath RC, Schulte-Merker S, Makinen T, Vikkula M, Mortimer PS, Mansour S, Jeffery S. Mutations in KIF11 cause autosomal-dominant microcephaly variably associated with congenital lymphoedema and chorioretinopathy. Am J Hum Genet 2012, 90(2) :356-62. IF=10.603
Butler MG, Dagenais SL, Garcia-Perez JL, Brouillard P, Vikkula M, Strouse P, Innis JW, Glover TW. Microcephaly, intellectual impairment, bilateral vesicoureteral reflux, distichiasis, and glomuvenous malformations associated with a 16q24.3 contiguous gene deletion and a glomulin mutation. Am J Med Genet part A 2012, 158A(4) :839-49. IF=2.304
Pietilä R, Nätynki M, Tammela T, Kangas J, Pulkki KH, Limaye N, Vikkula M, Koh GY, Saharinen P, Alitalo K, Eklund L. Ligand oligomerization state controls Tie2 receptor trafficking and Angiopoietin-2 ligand-specific responses. J Cell Sci 2012, 125(Pt 9) :2212-23. IF=6.111
Foulquier F*, Amyere M*, Jaeken J, Zeevaert R, Schollen E, Race V, Bammens R, Morelle W, Rosnoblet C, Legrand D, Demaegd D, Buist N, Cheillan D, Guffon N, Morsomme P, Annaert W, Hudson F, Van Schaftingen E, Vikkula M, Matthijs G. TMEM165 deficiency causes a congenital disorder of glycosylation. Am J Hum Genet 2012, 91 :15-26. *These authors contributed equally to this work. IF=10.603
Jaureguiberry G*, De la Dure-Molla M*, Parry D*, Quentric M*, Himmerkus N*, Koike T*, Poulter J, Klootwijk E, Robinette SL, Howie AJ, Patel V, Figueres M-L, Stanescu HC, Issler N, Nicholson JK, Bockenhauer D, Laing C,

Walsh SB, McCredie DA, Povey S, Asselin A, Picard A, Coulomb A, Medlar AJ, Bailleul-Forestier I, Verloes A, Le Caignec C, Roussey G, Guiol J, Isidor B, Logan C, Shore R, Johnson C, Inglehearn C, Al-Bahlani S, Schmittbuhl M, Clauss F, Huckert M, Laugel V, Ginglinger E, Pajarola S, Spartà G, Bartholdi D, Rauch A, Addor M-C, Yamaguti PM, Safatle HP, Acevedo AC, Martelli-Júnior H, dos Santos Netos PE, Coletta RD, Gruessel S, Sandmann C, Ruehmann D, Langman CB, Scheinman SJ, Ozdemir-Ozenen D, Hart TC, Hart PS, Neugebauer U, Schlatter E, Houillier P, Gahl WA, Vikkula M, Bloch-Zupan A, Bleich M, Kitagawa H, Unwin RJ, Mighell A, Berdal A, Kleta R. Nephrocalcinosis (Enamel Renal Syndrome) caused by autosomal recessive FAM20A mutations. Nephron Physiol 2012, 122(1-2) :1-6. *These authors contributed equally to this work. IF=1.571

Alders M, Mendola A, Adès L, Al Gazali L, Bellini C, Dallapiccola B, Edery P, Frank U, Hornshuh F, Huisman SA, Jagadeesh S, Kayserili H, Keng WT, Lev D, Prada CE, Sampson JR, Schmidtke J, Shashi V, van Bever Y, Van der Aa N, Verhagen JM, Verheij JB, Vikkula M, Hennekam RC. Evaluation of clinical manifestations in patients with severe lymphedema with and without CCBE1 mutations. Mol Syndromol 2013, 4(3):107-13.

Gianfrancesco F, Rendina D, Merlotti D, Esposito T, Amyere M, Formicola D, Muscariello R, De Filippo G, Strazzullo P, Nuti R, Vikkula M, Gennari L. Giant cell tumor occuring in familial Paget's disease of bone: report of clinical characteristics and linkage analysis of a large pedigree. J Bone Miner Res 2013, 28(2):341-350. IF=6.589

D'Auria L, Fenaux M, Van Der Smissen P, Chantrain C, Vermylen C, Vikkula M, Courtoy PJ, Tyteca D. Micrometric segregation of fluorescent membrane lipids: relevance for endogenous lipids and biogenesis in erythrocytes. J Lipid Res 2013, 54(4):1066-76. IF=4.730

Lannoy N, Grisart B, Eeckhoudt S, Verellen-Dumoulin C, Lambert C, Vikkula M, Hermans C. Intron 22 homologous regions are implicated in exons 1-22 duplications of the F8 gene. Eur J Hum Genet 2013, 21(9):970-6. IF=4.400

Ballieux F, Modarressi A, Hammer F, Clapuyt Ph, Docquier P-L, Godfraind C, Feyaerts A, Brouillard P, Vikkula M, Boon LM. Reconstructive surgery in the management of a patient with CLOVES Syndrome. JPRAS 2013, 66(12):1813-5. IF=1.474

De la Dure-Molla M, Quentric M, Yamaguti PM, Acevedo A-C, Mighell AJ, Vikkula M, Huckert M, Berdal A, Bloch-Zupan A. Pathognomonic oral profile of enamel renal syndrome (ERS) caused by receessive FAM20A mutations. Orphanet J Rare Dis 2014, 9 :84. IF=3.358

Rauen KA *, Huson SM, Burkitt-Wright E, Evans GD, Farschtschi S, Ferner S, Gutmann DH, Hanemann O, Kerr B, Legius E, Parada L, Patton M, Peltonen J, Ratner N, Riccardi VM, Van der Vaart T, Vikkula M, Viskochil DH, Zenker M, Upadhyaya M. Recent developments in neurofibromatoses and RASopathies: management, diagnosis and current and future therapeutic avenues. Am J Med Genet part A 2015, 167(1) :1-10. IF2014=2.159

Moalem S, Brouillard P, Kuypers D, Legius E, Harvey E, Taylor G, François M, Vikkula M, Chitayat D. Hypothrichosis-Lymphedema-Telangiectasia Renal defect associated with a truncating mutation in the SOX18 gene. Clin Genet 2015, 87(4):378-82. IF=3.89

Papathomas TG, Oudijk L, Persu A, Gill A, van Nederveen F, Tischler A, Tissier F, Volante M, Matias-Guiu X, Smid M, Favier J, Rapizzi E, Libe R, Currás-Freixes M, Aydin S, Huynh T, Lichtenauer U, van Berkel A, Canu L, Domingues R, Clifton-Bligh R, Bialas M, Vikkula M, Baretton G, Papotti M, Nesi G, Badoual C, Pacak K, Eisenhofer G, Timmers H, Beuschlein F, Bertherat J, Mannelli M, Robledo M, Gimenez-Roqueplo A-P, Dinjens W, Korpershoek E, de Krijger RR. SDHB/SDHA immunohistochemistry in pheochromocytomas and paragangliomas: a multicenter interobserver variation analysis using virtual microscopy. A Multinational Study of the European Network for the Study of Adrenal Tumours (ENS@T). Mod Pathol 2015, 28(6) :807-21. IF2014=6.187

Troilius A, Lauritzen E, Ljunggren B, Revencu N, Vikkula M, Svensson A. Heredity of port-wine stains: investigation of families without a RASA1 mutation. J Cosm & Laser Therapy 2015, 17(4) :204-8.

Wassef M, Blei F, Adams DM, Alomari A, Baselga E, Berenstein A, Burrows P, Frieden I, Garzon M, Lopez-Gutierrez J-C, Lord D, Mitchel S, Powell J, Prendiville J, Vikkula M ; ISSVA Board and Scientific Committee. Vascular Anomalies Classification: Recommendations from the International Society for The Study of Vascular Anomalies. Pediatrics 2015, 136(1) :e203-e214. IF2014=5.473

Lannoy N, Lambert C, Vikkula M, Hermans C. Overrepresentation of missense mutations in mild hemophilia A patients from Belgium: founder effect or independent occurrence ? Thromb Res 2015, 135 :1057-63. IF2014=2.447

Boudon E, Levy T, Abossolo T, Cartault F, Brouillard P, Vikkula M, Kieffer-Traversier M, Ramful D, Alessandri JL. Antenatal presentation of hereditary lymphedema type I. Eur J Med Genet 2015, 58(6-7) :329-31. IF=1.81

Lannoy N, Bandelier C, Grisart B, Reginster M, Ronge-Collard E, Vikkula M, Hermans C. Tandem inversion duplication within F8 intron 1 associated with mild haemophilia A. Haemophilia 2015, 21(4) :516-22. IF2014=2.603

Nätynki M, Kangas J, Miinalainen I, Sormunen R, Pietilä R, Soblet J, Boon LM, Vikkula M, Limaye N, Eklund L. Common and specific effects of TIE2 mutations causing venous malformations. Hum Mol Genet 2015 Nov 15, 24(22) :6374-89. IF=5.99

Evenepoel L, Van Nederveen FH, Oudijk L, Papathomas TG, Restuccia DF, Belt EJ, Franssen GJ, Feelders RA, Van Eeden S, Timmers H, De Herder WW, Aydin S, Vikkula M, De Krijger RR, Dinjens WN, Persu A, Korpershoek E. 9B.09: Identification of markers predictive for malignant behavior of pheochromocytomas and paragangliomas. J Hypertens 2015 Jun;33 Suppl 1:e122. doi: 10.1097/01.hjh.0000467681.47178.d5.

Christiaens AB, Deprez PM, Amyere M, Mendola A, Bernard P, Gillerot Y, Clapuyt P, Godfraind C, Lengelé BG, Vikkula M, Nyssen-Behets C. Isolated bilateral transverse agenesis of the distal segments of the lower limbs at the level of the knee joint in a human fetus. Am J Med Genet A. 2016 170(2) :523-30.

Lannoy N, Ravoet M, Grisart B, Fretigny M, Vikkula M, Hermans C. Five int22h homologous copies at the Xq28 locus identified in intron22 inversion type 3 of the Factor VIII gene. Thromb Res 2016, 137 :224-7.

Havelange V, Ameye G, Théate I, Callet-Bauchu E, Lippert E, Luquet I, Raphaël M, Vikkula M, Poirel HA. The peculiar 11q-gain/loss aberration reported in a subset of MYC-negative high-grade B-cell lymphomas can also occur in a MYC-rearranged lymphoma. Cancer Genet 2015, 209(3) :117-8.

Havelange V, Pepermans X, Ameye G, Théate I, Callet-Bauchu E, Barin C, Penther D, Lippert E, Michaux L, Mugneret F, Dastugue N, Raphaël M, Vikkula M, Poirel HA. Genetic differences between paediatric and adult Burkitt lymphomas. Br J Haematol 2016, 173(1) :137-44.

Persu A, Evenepoel L, Jin Y, Mendola A, Ngueta G, Yang W-Y, Gruson D, Horman S, Staessen JA, Vikkula M. STK39 and WNK1 are potential hypertension susceptibility genes in the BELHYPGEN cohort. Medicine (Baltimore) 2016, 95(15) :e2968.

Goffinet L, Oussalah A, Guéant-Rodriguez RM, Chery C, Basha M, Avogbe PH, Josse T, Jeannesson E, Rouyer P, Flayac J, Gerard P, Le Touze A, Bonin-Goga B, Goga D, Simon E, Feillet F, Vikkula M, Guéant JL. Cystathionine β-synthase genetic variant rs2124459 is associated with a reduced risk of cleft palate in French and Belgian populations. J Med Genet 2016, 53(12) :828-34.

Castrén E, Salminen P, Vikkula M, Pitkäranta A, Klockars T. Inheritance patterns of Infantile Hemangioma. Pediatrics 2016, 138(5). IF=5.80

van Marcke C, De Leener A, Berlière M, Vikkula M, Duhoux FP. Routine use of gene panel testing in hereditary breast cancer should be performed with caution. In: Critical Reviews in Oncology / Hematology 2016, 108 : 33-9.

Jha KS, Rauniyar K, Karpanen T, Leppanen V-M, Brouillard P, Vikkula M, Alitalo K, Jeltsch M. Efficient activation of the lymphangiogenic growth factor VEGF-C requires the C- terminal domain of VEGF-C and the N-terminal domain of CCBE1. Sci Reports 2017,7(1) :4916.

Evenepoel L, Helaers R, Vroonen L, Aydin S, Hamoir M, Maiter D, Vikkula M, Persu P. KIF1B and NF1 are the most frequently mutated genes in paraganglioma and pheochromocytoma tumors. Endocr Relat Cancer 2017, 24(8) :L57-61.

Brambila-Tapia AJL, García-Ortiz JE, Brouillard P, Nguyen H-L, Vikkula M, Ríos-González BE, de Jesús Sandoval-Muñiz R, Sandoval-Talamantes AK, Bobadilla-Morales L, Corona-Rivera JR, Arnaud-Lopez L. GATA2 null mutation associated with incomplete penetrance in a family with Emberger syndrome. Hematology 2017, 22(8) :467-71.

Arts FA, Sciot R, Brichard B, Renard M, Noël LA, Velghe AI, Galant C, Debiec-Rychter M, Van Damme A, Vikkula M, Helaers R, Limaye N, A. Poirel H, Demoulin J-B. PDGFRB gain-of-function mutations in sporadic infantile myofibromatosis. Hum Mol Genet 2017, 26(10) :1801-10.

van Marcke C, Vikkula M, Duhoux FP. Comment on "Increased identification of candidates for high-risk breast cancer screening through expanded Genetic Testing". J Am Coll Radiol 2017 May;14(5):582.

Seront E, Vikkula M, Boon LM. Venous malformations. Otolaryngologic Clinics of North America 2018, 51(1) :173-84.

Evenepoel L, van Nederveen FH, Oudijk L, Papathomas TG, Restuccia DF, Belt EJT, Franssen GJH, Feelders RA, Perren A, van Eeden S, Timmers H, de Herder WW, Vroonen L, Aydin S, Vikkula M, de Krijger RR, Dinjens WNM,

Persu A*, Korpershoek E*. Expression of CNTN4 (contactin4) is associated with malignant behavior in pheochromocytomas and paragangliomas. J Clin Endocrin Metab 2018, 103(1) :46-55. Prix BES de la publication Belge 2017-2018.
Di Monaco S, Georges A, Lengelé J-Ph, Vikkula M, Persu A. Genomics of fibromuscular dysplasia. Intl J Mol Sci 2018, 19(5). pii :E1526
Boon LM, Vikkula M, Seront E. Venous malformations. Moise Levy, in press.
Laterre M, Bernard P, Vikkula M, Sznajer Y. Improved diagnosis in nonimmune hydrops fetalis using a standardized algorithm. Prenat Diagn 2018, 38 :337-43.
Van Marcke C, Collard A, Vikkula M, Duhoux F. Prevalence of pathogenic variants and variants of unknown significance in patients at high risk of breast cancer: a systematic review and meta-analysis of gene-panel data. Crit Rev Oncol Hematol 2018, 132 :138-44.
Hammer J, Seront E, Duez S, Dupont S, Van Damme A, Schmitz S, Hoyoux C, Chopinet C, Clapuyt Ph, Hammer F, Vikkula M, Boon LM. Sirolimus is efficacious in treatment for extensive and/or complex slow-flow vascular malformations: a monocentric prospective phase II study. Orphanet J Rare Dis 2018, 13(1) :191. IF=3.61
Janssens P, Dekeuleneer V, Van Damme A, Brouillad P, Revencu N, Clapuyt Ph, Ferreira I, Ballieux F, Vikkula M, Marot L, Baeck M, Boon LM. Angiosarcoma arising from congenital primary lymphedema. Ped Dermatol 2018, 35 :e382-8.
Narasimhaiah D, Legrand C, Remark R, Damotte D, Munda M, De Potter P, Coulie PG, Vikkula M, Godfraind C. DNA alteration-based classification of uveal melanoma gives better prognostic stratification than immune infiltration, which has a neutral effect in the high-risk group. Cancer Med. 2019 Jun;8(6):3036-3046.
Ciurica S, Lopez-Sublet M, Loeys BL, Radhouani I, Natarajan N, Vikkula M, Maas AHEM, Adlam D, Persu A. Arterial tortuosity: novel implications for an old phenotype. Hypertension 2019, 73(5) :951-60.
Dachy G, de Krijger RR, Fraitag S, Théate I, Brichard B, Hoffman SB, Libbrecht L, Arts FA, PALGA group, Brouillard P, Vikkula M, Limaye N, Demoulin JB. Association of PDGFRB mutations with pediatric myofibroma and myofibromatosis. JAMA Dermatol 2019, Apr 24. doi: 10.1001/jamadermatol.2019.0114
Rauen KA, …Vikkula M, Widemann BC, Wolkenstein P, Upadhyaya M. First international conference on RASopathies and Neurofibromatosis in Asia: identification and advances of new therapeutics. Am J Med Genet Part A 2019, Jun;179(6):1091-1097.
Duran D, Zeng X, Jin SC, Choi J, Nelson-Williams C, Yatsula B, Gaillard J, Furey CG, Lu Q, Timberlake AT, Dong W, Sorscher MA, Loring E, Klein J, Allocco A, Hunt A, Conine S, Karimy JK, Youngblood MW, Zhang J, DiLuna ML, Matouk CC, Mane S, Tikhonova IR, Castaldi C, Lopez-Giraldez F, Knight J, Haider S, Soban M, Alper SL, Komiyama M, Ducruet AF, Zabramski JM, Dardik A, Walcott BP, Stapleton CJ, Aagaard-Kienitz B, Rodesch G, Jackson E, Smith ER, Orbach DB, Berenstein A, Bilguvar K, Vikkula M, Gunel M, Lifton RP, Kahle KT. Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation. Neuron 2019 Feb 6;101(3):429-443.e4.
Roux A, Vikkula M, Pallud J. Is developmental venous anomaly an imaging biomarker of PIK3CA mutated gliomas ? Neurosurgery 2020, 86(1):E93.
Lobeek D, Bouwman FCM, Aarntzen EHJG, Molkenboer-Kuenen JDM, Flucke UE, Nguyen HL, Vikkula M, Boon LM, Klein W, Laverman P, Oyen WJG, Boerman OC, Terry SYA, SchultzeKool LJ, Rijpkema M. A clinical feasibility study to Image angiogenesis in patients with arteriovenous malformations using 68Ga-RGD PET/CT. J Nucl Med. 2020, 61(2):270-5.
Iznardo H, Roé E, Puig L, Vikkula M, Lopez-Sanchez C, Baselaga E. Good response to pulsed dye laser in patients with capillary malformation-arteriovenous malformation syndrome (CM-AVM). Pediatr Dermatol 2020, 37(2):342-344.
Martinez-Corral I, Zhang Y, Petkova M, Ortsäter H, Sjöberg S, Castillo SD, Brouillard P, Libbrecht L, Saur D, Graupera M, Alitalo K, Boon LM, Vikkula M, Mäkinen T. Blockade of VEGF-C signaling inhibits lymphatic malformations driven by oncogenic Pik3ca mutation. Nat Commun 2020, 11(1):2869.
Galot R, van Marcke C, Helaers R, Mendola A, Goebbels RM, Caignet X, Ambroise J, Wittouck K, Vikkula M, Limaye N, Machiels JH. Liquid biopsy for mutational profiling of locoregional recurrent and/or metastatic head and neck squamous cell carcinoma. Oral Oncol 2020 Mar 10;104:104631.
Van Marcke C, Helaers R, De Leener A, Merhi A, Schoonjans CA, Ambroise J, Galant C, Delrée P, Rothé F, Bar I, Khoury E, Brouillard P, Canon J-L, Vuylsteke P, Machiels J-P, Berlière M, Limaye N, Vikkula M, Duhoux F. Tumor sequencing is useful to refine the analysis of germline variants in unexplained high-risk breast cancer families. Breast Cancer Res 2020, 22(1):36.
Verstraeten A, Perik M, Baranowska A, Meester J, Van Den Heuvel L, Bastianen J, Kempers M, Krapels IPC, Maas A, Rideout A, Vandersteen A, Sobey G, Johnson D, Fransen E, Ghali N, Webb T, Al-Hussaini A, de Leeuw P, Delmotte Ph, Lopez-Sublet M, Pappaccogli M, Sprynger M, Toubiana L, Van Laer L, van Dijk F, Vikkula M, Samani N, Persu A, Adlam D, Loeys B. Enrichment of rare variants in Loeys-Dietz syndrome genes in spontaneous coronary artery dissection but not in severe fibromuscular dysplasia. Circulation 2020, 142(10):1021-1024.
Boucinha C, Caetano AR, Santos HL, Helaers R, Vikkula M, Branquinha MH, Dos Santos ALS, Grellier P, Morelli KA, d'Avila-Levy CM. Analysing ambiguities in trypanosomatids taxonomy by barcoding. Mem Inst Oswaldo Cruz. 2020 Jun 19;115:e200504.
Hansmann P, Brückner A, Kiontke S, Berkenfeld B, Seebohm G, Brouillard P, Vikkula M, Jansen FE, Nellist M, Oeckinghaus A, Kümmel D. Structure of the TSC2 GAP domain: mechanistic insight into catalysis and pathogenic mutations. Structure 2020.
Pollet H, Cloos A-S, Stommen A, Conrard L, Paquot A, Ghodsi M, Vanderroost J, Carquin M, Leonard C, Guthmann M, Lingurski M, Vermylen Ch, Killian Th, Gatto L, Rider M, Pyr Sdit Ruys, Vertommen D, Vikkula M, Brouillard P, Van Der Smissen P, Muccioli G, Tyteca D. Aberrant membrane composition and biophysical properties impair erythrocyte morphology and functionality in elliptocytosis. Biomolecules 2020, 10(8):1120.
Gerotziafas GT, Catalano M, Colgan MP, Pecsvarady Z, Wautrecht JC, Fazeli B, Olinic DM, Farkas K, Elalamy I, Falanga A, Fareed J, Papageorgiou C, Arellano RS, Agathagelou P, Antic D, Auad L, Banfic L, Bartolomew JR, Benczur B, Bernardo MB, Boccardo F, Cifkova R, Cosmi B, De Marchi S, Dimakakos E, Dimopoulos MA, Dimitrov G, Durand-Zaleski I, Edmonds M, El Nazar EA, Erer D, Esponda OL, Gresele P, Gschwandtner M, Gu Y, Heinzmann M, Hamburg NM, Hamadé A, Jatoi NA, Karahan O, Karetova D, Karplus T, Klein-Weigel P, Kolossvary E, Kozak M, Lefkou E, Lessiani G, Liew A, Marcoccia A, Marshang P, Marakomichelakis G, Matuska J, Moraglia L, Pillon S, Poredos P, Prior M, Salvador DRK, Schlager O, Schernthaner G, Sieron A, Spaak J, Spyropoulos A, Sprynger M, Suput D, Stanek A, Stvrtinova V, Szuba A, Tafur A, Vandreden P, Vardas PE, Vasic D, Vikkula M, Wennberg P, Zhai Z; Scientific Reviewer Committee. Guidance for the Management of Patients with Vascular Disease or Cardiovascular Risk Factors and COVID-19: Position Paper from VAS-European Independent Foundation in Angiology/Vascular Medicine. Thromb Haemost. 2020 Dec;120(12):1597-1628.
Persu A, Vikkula M, Loeys B. PTGIR, a susceptibility gene for fibromuscular dysplasia? Cardiovasc Res. 2021 Mar 21;117(4):990-992.
Mäkinen T, Boon LM, Vikkula M, Alitalo K. Lymphatic Malformations: Genetics, Mechanisms and Therapeutic Strategies. Circ Res. 2021 Jun 25;129(1):136-154.
El-Sibai M, El Hajj J, Al Haddad M, El Baba N, Al Saneh M, Daoud Khatoun W, Helaers R, Vikkula M, El Atat O, Sabbagh J, Abou Chebel N, Ghassibe-Sabbagh M. Dysregulation of Rho GTPases in orofacial cleft patients-derived primary cells leads to impaired cell migration, a potential cause of cleft/lip palate development. Cells Dev. 2021 Mar;165:203656.
Georges A, Yang ML, Berrandou TE, Bakker MK, Dikilitas O, Kiando SR, Ma L, Satterfield BA, Sengupta S, Yu M, Deleuze JF, Dupré D, Hunker KL, Kyryachenko S, Liu L, Sayoud-Sadeg I, Amar L, Brummett CM, Coleman DM, d'Escamard V, de Leeuw P, Fendrikova-Mahlay N, Kadian-Dodov D, Li JZ, Lorthioir A, Pappaccogli M, Prejbisz A, Smigielski W, Stanley JC, Zawistowski M, Zhou X, Zöllner S; FEIRI investigators; International Stroke Genetics Consortium (ISGC) Intracranial Aneurysm Working Group; MEGASTROKE, Amouyel P, De Buyzere ML, Debette S, Dobrowolski P, Drygas W, Gornik HL, Olin JW, Piwonski J, Rietzschel ER, Ruigrok YM, Vikkula M, Warchol Celinska E, Januszewicz A, Kullo IJ, Azizi M, Jeunemaitre X, Persu A, Kovacic JC, Ganesh SK, Bouatia-Naji N. Genetic investigation of fibromuscular dysplasia identifies risk loci and shared genetics with common cardiovascular diseases. Nat Commun. 2021 Oct 15;12(1):6031.
Latosinska A, Bruno RM, Pappaccogli M, Bacca A, Beauloye C, Boutouyrie P, Khettab H, Staessen JA, Taddei S, Toubiana L, Vikkula M, Mischak H, Persu A. Increased Collagen Turnover Is a Feature of Fibromuscular Dysplasia and

Associated With Hypertrophic Radial Remodeling: A Pilot, Urine Proteomic Study. Hypertension. 2022 Jan;79(1):93-103.
Homayun-Sepehr N*, McCarter AL*, Helaers R, Galant C, Boon LM, Brouillard P, Vikkula M*, Dellinger MT*. KRAS-driven model of Gorham-Stout disease effectively treated with trametinib. JCI Insight. 2021 Aug 9;6(15):e149831. (*Underlined authors contributed equally to this work).
Byrne AB*, Brouillard P*, Sutton DL*, Kazenwadel J*, Montazaribarforoushi S, Secker GA, Oszmiana A, Babic M, Betterman KL, Brautigan PJ, White M, Piltz SG, Thomas PQ, Hahn CN, Rath M, Felbor U, Korenke GC, Smith CL, Wood KH, Sheppard SE, Adams DM, Kariminejad A, Helaers R, Boon LM, Revencu N, Moore L, Barnett C, Haan E, Arts P, Vikkula M, Scott HS, Harvey NL. Pathogenic variants in MDFIC cause recessive central conducting lymphatic anomaly with lymphedema. Sci Transl Med. 2022 Mar 2;14(634): eabm4869. (*Underlined authors contributed equally to this work).
Tas B, Starnoni D, Smajda S, Vivanti AJ, Adamsbaum C, Eyries M, Melki J, Tawk M, Ozanne A, Revencu N, Soubrier F, Siala S, Vikkula M, Deiva K, Saliou G. Arteriovenous Cerebral High Flow Shunts in Children: From Genotype to Phenotype. Front Pediatr. 2022 Apr 25.
Dompmartin A, van der Vleuten CJM, Dekeuleneer V, Duprez T, Revencu N, Désir J, Te Loo DMWM, Flucke U, Eijkelenboom A, Schultze Kool L, Vikkula M, Boon L. GNA11-mutated Sturge-Weber syndrome has distinct neurological and dermatological features. Eur J Neurol. 2022 Oct;29(10):3061-3070.
Papadimitriou S, Gravel B, Nachtegael C, De Baere E, Loeys B, Vikkula M, Smits G, Lenaerts T. Toward reporting standards for the pathogenicity of variant combinations involved in multilocus/oligogenic diseases. HGG Adv. 2022 Dec 2;4(1):100165.
Cloos AS, Pollet H, Stommen A, Maja M, Lingurski M, Brichard B, Lambert C, Henriet P, Pierreux C, Pyr Dit Ruys S, Van Der Smissen P, Vikkula M, Gatto L, Martin M, Brouillard P, Vertommen D, Tyteca D. Splenectomy improves erythrocyte functionality in spherocytosis based on septin abundance but not maturation defects. Blood Adv. 2023 Feb 8:bloodadvances.2022009114.
Guilhem A, Dupuis-Girod S, Espitia O, Rivière S, Seguier J, Kerjouan M, Lavigne C, Maillard H, Magro P, Alric L, Lipsker D, Parrot A, Leguy V, Vanlemmens C, Guibaud L, Vikkula M, Eyries M, Valette PJ, Giraud S. Seven cases of hereditary haemorrhagic telangiectasia-like hepatic vascular abnormalities associated with EPHB4 pathogenic variants. J Med Genet. 2023 Feb 22:jmg-2022-109107.
Afshar Y, Ma F, Quach A, Jeong A, Sunshine HL, Freitas V, Jami-Alahmadi Y, Helaers R, Li X, Pellegrini M, Wohlschlegel JA, Romanoski CE, Vikkula M, Iruela-Arispe ML. Transcriptional drifts associated with environmental changes in endothelial cells. Elife. 2023 Mar 27;12
Seront, E., Biard, J.M., Van Damme, A., Revencu N., Lengelé B., Schmitz S., de Toeuf C, Clapuyt, P., Veyckemans, F., Prégardien C., Vikkula M., Bernard P., Boon, LM. A case report of sirolimus use in early fetal management of lymphatic malformation. Nat Cardiovasc Res 2, 595–599 (2023).
Seront E, Van Damme A, Legrand C, Bisdorff-Bresson A, Orcel P, Funck-Brentano T, Pietri MA, Dompmartin A, Quere I, Brouillard P, Revencu, N, De Bortoli M, Hammer F, .Clapuyt P, Dumitriu D, Vikkula M, Boon L, Preliminary Results of the European Multicentric Phase III Trial regarding Sirolimus in Slow-Flow Vascular Malformations. JCI Insight. 2023;8(21):e173095.
Atique Tacla M, de Mello Copelli M, Pairet E, Lopes Monlleó I, Marques Ribeiro E, Lustosa Mendes E, Helaers R, Paiva Vieira T, Vikkula M, Gil-da-Silva-Lopes V. Molecular investigation in individuals with orofacial clefts and microphthalmia-anophthalmia-coloboma spectrum. Eur J Hum Genet (2023).
Tuleja A, Bernhard S, Hamvas G, Andreoti TA, Rössler J, Boon L, Vikkula M, Kammer R, Haupt F, Döring Y, Baumgartner I. Clinical phenotype of adolescent and adult patients with extracranial vascular malformation. J Vasc Surg Venous Lymphat Disord. 2023 Sep;11(5):1034-1044.e3.
Guilhem A, Dupuis-Girod S, Espitia O, Rivière S, Seguier J, Kerjouan M, Lavigne C, Maillard H, Magro P, Alric L, Lipsker D, Parrot A, Leguy V, Vanlemmens C, Guibaud L, Vikkula M, Eyries M, Valette PJ, Giraud S. Seven cases of hereditary haemorrhagic telangiectasia-like hepatic vascular abnormalities associated with EPHB4 pathogenic variants. J Med Genet. 2023 Sep;60(9):905-909.
Lagnaux E, Boon LM, Revencu N, Vikkula M, Hermans C. Direct oral anticoagulants and venous malformations: literature review and retrospective study of 29 patients. Research and Practice in Thrombosis and Haemostasis. March 2024. 8(3).
Seront E, Froidure A, Revencu N, Dekeuleneer V, Clapuyt P, Dumitriu D, Vikkula M, Boon LM. Targeted treatment in complex lymphatic anomaly: a case of synergistic efficacy of trametinib and sirolimus. Orphanet J Rare Dis. 2024 May 16;19(1):199.
Nguyen, H. L., Holderbaum Do Amaral, R., Lerouge, S., De Roo, A. K., Zehtabi, F., Vikkula, M., & Soulez, G. (2024). Injectable chitosan hydrogel effectively controls lesion growth in a venous malformation murine model. Diagnostic and interventional imaging, Nov;105(11):430-438. doi: 10.1016/j.diii.2024.07.004. Epub 2024 Aug 2. PMID: 39095271.
Serra, M., Rediti, M., Collet, L., Lifrange, F., Venet, D., Occelli, N., Wang, X., Vincent, D., Rouas, G., Craciun, L., Larsimont, D., Buisseret, L., Vikkula, M., Duhoux, F. P., Rothé, F., & Sotiriou, C. (2024). 113MO Spatial transcriptomics-derived classification of invasive lobular carcinoma: Associations with clinical, genomic characteristics, and prognosis. ESMO Open, 9.
Karakose, B., Serra, M., van Renne, N., Lifrange, F., Venet, D., Vincent, D., Rouas, G., Rediti, M., Craciun, L., Larsimont, D., Vikkula, M., Duhoux, F. P., Rothé, F., & Sotiriou, C. (2024). 166P Unravelling HR-positive, HER2-negative breast cancer and its tumor microenvironment heterogeneity using spatial transcriptomics. ESMO Open, 9.
Descos, M., Leaute-Labreze, C., Vikkula, M., Delfour, C., Bigorre, M., Kuentz, P., & Bessis, D. (2024). Malformation capillaire segmentaire avec atrophie centrale, une nouvelle forme clinique de mosaïcisme somatique p.Gln209Pro GNA11. Annales de Dermatologie et de Vénéréologie - FMC, 4(8, Supplement 1), A208-A209.
Andreoti, T.-A.A., Maiolo, M., Tuleja, A., Döring, Y., Schaller, A., Vassella, E., Boon, L.M., Baumgartner, I., Bernhard, S.M., Zweier, C., Vikkula, M. and Rössler, J. (2025), Non-Hotspot PIK3CA Variants Have Higher Variant Allele Frequency and are More Common in Syndromic Vascular Malformations. Am J Med Genet, 197: e63883.
Holm, A., Graus, M. S., Wylie-Sears, J., Tan, J. W. H., Alvarez-Harmon, M., Borgelt, L., Nasim, S., Chung, L., Jain, A., Sun, M., Sun, L., Brouillard, P., Lekwuttikarn, R., Qi, Y., Teng, J., Vikkula, M., Kozakewich, H., Mulliken, J. B., Francois, M., & Bischoff, J. (2025). An endothelial SOX18–mevalonate pathway axis enables repurposing of statins for infantile hemangioma. The Journal of clinical investigation, 135(7).
Gucciardo, F., Lebeau, A., Pirson, S., Buntinx, F., Ivanova, E., Blacher, S., Brouillard, P., Deroye, J., Baudin, L., Pirnay, A., Morfoisse, F., Villette, C., Nizet, C., Lallemand, F., Munaut, C., Alitalo, K., Geris, L., Vikkula, M., Gautier-Isola, M., & Noel, A. Targeting uPARAP Modifies Lymphatic Vessel Architecture and Attenuates Lymphedema. Circulation, 0(0).
Coulie, J., Seront, E., Vikkula, M., & Boon, L. M. (2025). Extracranial arteriovenous malformations: towards etiology-based therapeutic management. The Journal of clinical investigation, 135(6).
Seront E, Coulie J, Dekeuleneer V, Hammer F, Vikkula M, Boon LM. Monocentric pilot trial of trametinib in severe extracranial arteriovenous malformations. J Clin Oncol 43, 3096-3096(2025).
Seront E, Damme A, Coulie J, Vikkula M, Boon LM. Personalized sirolimus regimen for vascular malformations: a retrospective analysis of vase cohort. Orphanet J Rare Dis. 2025.
Copelli, M. d. M., Atique-Tacla, M., Pairet, E., Correia-Costa, G. R., Henrique de Souza, T., Monlleó, I. L., Vieira, T. P., Helaers, R., Vikkula, M., & Gil-da-Silva-Lopes, V. L. (2025). Whole exome sequencing in 18 Brazilian families with vertical transmission of non-syndromic oral clefts. Journal of Cranio-Maxillofacial Surgery, 53(4), 370-376.
Martin-Valiente, E., Du, Y., Goemans, C., America, M., Zindy, E., Adam, M., Scheid, B., Vikkula, M., Lubicz, B., Vanhollebeke, B., & Baeyens, N. (2025). Reversal of cerebrovascular anomalies in a zebrafish model of vein of Galen aneurysm. Nature cardiovascular research, 4(6), 773–789.
Coulie, J., Vikkula, M., & Boon, L. M. (2025). Peripheral arteriovenous malformations: diagnosis and future prospects. Presse medicale (Paris, France : 1983), 104296. Advance online publication.
Goldenberg, D.C, Vikkula, M., Penington, A., Blei, F., Schultze-Kool, L., Wassef, M., Frieden, I.J. the ISSVA Vascular Anomalies Classification Group. Updated Classification of Vascular Anomalies: A living document from the International Society for the Study of Vascular Anomalies Classification Group. Journal of Vascular Anomalies 6(2):p e113, June 2025.
Kraft, M., Schoofs, H., Petkova, M., Andrade, J., Grosso, A. R., Benedito, R., De Roo, A. K., Boon, L. M., Vikkula, M.,

Kapp, F. G., Hägerling, R., Potente, M., & Mäkinen, T. (2025). Angiopoietin-TIE2 feedforward circuit promotes PIK3CA-driven venous malformations. Nature cardiovascular research. Nat Cardiovasc Res 4, 801–820 (2025).
Tuleja A., Döring Y., Helfenstein F.N., Andreoti T-A., Rössler J., Boon L.M., Vikkula M., Haupt F., Hamvas G., Bernhard S.M., Baumgartner I. Alcohol embolization versus non-invasive treatment for pain relief in peripheral venous malformations: a comparative study. Cardiovasc Diagn Ther 2025;15(3):598-609
Karakose B, Serra M, Van Renne N, Lifrange F, Venet D, Vincent D, Rouas G, Rediti M, Craciun L, Larsimont D, Vikkula M, Duhoux FP, Rothé F, Sotiriou C. 159P Identification of 4 molecular HR+/HER2- breast cancer subtypes with distinct biological and clinical behaviors using spatial transcriptomics. 2025. Annals of Oncology, Volume 36, S269.
Serra M, Rediti M, Collet L, Lifrange F, Venet D, Occelli N, Papagiannis A, Vincent D, Rouas G, Larsimont D, Vikkula M, Duhoux FP, Rothé F, Sotiriou C. Spatial transcriptomics reveals tumor microenvironment-driven subtypes of invasive lobular carcinoma. Proc Natl Acad Sci U S A. 2026 Feb 10;123(6):e2517567123

C. Chapters & Reviews

Vikkula M, Olsen BR. Unravelling the molecular genetics of osteoarthrosis. Ann Med. Aug 1996;28(4):301-4. IF=3.84
Vikkula M, Boon LM, Mulliken JB, Olsen BR. Molecular basis of vascular anomalies. Trends Cardiovasc Med. Oct 1998;8(7):281-292. IF=4.77
Karttunen L, Vikkula M, Boon LM, Olsen BR. TIE-2 signalling and venous malformations. In: Angiogenesis in health and disease: basic mechanisms and clinical applications. Gabor M. Rubanyi, ed., Marcel Dekker Inc. New York, NY, 1999, pp. 89-96.
Karttunen L, Felbor U, Vikkula M, Olsen BR. Vascular matrix and disorders. In: Hematopoiesis: A Developmental Approach. Leonard Zon, ed. Oxford University Press, New York, NY, 2001, pp. 784-795.
Vikkula M, Boon LM, Mulliken JB. Molecular genetics of vascular malformations. Matrix Biol Sep 2001;20(5-6):327-335. IF=4.47
Vikkula M. An update on Molecular Basis of Vascular Malformations. In: Benigne Gefäßfehl- und Neubildungen der Haut (Benign vascular malformations and tumors). By Michael Landthaler, Ulrich Hohenleutner and Thomas Vogt. Ed.: Blackwell Verlag GmbH, Berlin, Germany, 2002, pp. 15-24.
Brouillard P, Vikkula M. Vascular malformations: localized defects in vascular morphogenesis. Clin Genet. May 2003;63(5):340-51. IF=3.28
Vikkula M, Karkkainen MJ, Alitalo K. Angiopoietins, TIEs, Ephrins, VEGFs and VEGFRs. In : Inborn errors of development: the molecular basis of clinical disorders on morphogenesis. Editors C.J. Epstein, R.P. Erickson, and A. Wynshaw-Boris, Oxford University Press, New York, NY, (2004), 987-996.
Vikkula M. Identification of implicated factors in angiogenesis for the study of vascular anomalies. Bull Mem Acad R Med Belg. 2005;160(10-12):435-443; discussion 443-434.
Brouillard P, Ghassibe M, Penington A, Boon LM, Dompmartin A, Temple K, Cordisco M, Adams D, Piette F, Harper J, Syed S, Boralevi F, Taïeb A, Danda S, Baselga E, Enjolras O, Mulliken JB, Vikkula M. Four common glomulin mutations cause two thirds of glomuvenous malformations ("familial glomangiomas"): evidence for a founder effect. J Med Genet. Feb 2005;42(2):e13. IF=4.33
Aerts V, Brouillard P, Boon LM, Vikkula M. Glomuvenous malformations. In: Atlas of Genetics and Cytogenetics in Oncology and Hematology. Editeur: Jean-Loup Huret, Genetics DIM, University Hospital, France. (http://atlasgeneticsoncology.org//Genes/GLMNID43022ch1p22.html)
Casanova D, Boon LM, Vikkula M. Venous malformations: clinical characteristics and differential diagnosis. Ann Chir Plast Esthet. Aug-Oct 2006;51(4-5):373-87.
Persu A, Vikkula M. SDH genes : from glomic tumours to pheocromocytomas. Curr Hypertens Rev 2006, 1 :267-73.
Revencu N, Vikkula M. Cerebral cavernous malformation: new molecular and clinical insights. J Med Genet. Sep 2006;43(9):716-21. IF=4.33
Ghassibe M, Bayet B, Revencu N, Desmyter L, Verellen-Dumoulin C, Gillerot Y, Deggouj N, Vanwijck R, Vikkula M. Orofacial clefting: update on the role of genetics. B-Ent 2006;2 Suppl 4:20-4.
Persu A, Vikkula M. Génétique et maladies cardiovasculaires athéromateuses. Athérosclérose Athérothrombose TransMed 2006, p. 191-209.
Pocock B, Boon LM, Vikkula M. Molecular basis of vascular birthmarks. Sem Plast Surg 2006, 20(3) :149-56.
Vikkula M. Pathogénie et génétique des anomalies vasculaires (Pathogenesis and genetics of vascular anomalies). Ann Chir Plast Esthet 2006 ;51(4-5) :282-6.
Vikkula M. Vascular anomalies. Exp Dermatol 2007; 16:860-79. IF=2.951; 0 citations.
Vikkula M. Vascular Pathologies: Angiogenomics: towards a genetic nosology and understanding of vascular anomalies. Eur J Hum Genet 2007, 15 :821-2. IF=3.697
Brouillard P and Vikkula M. Genetic causes of vascular malformations. Hum Mol Genet 2007, 16 :R140-9. IF=7.806; 105 citations.
Godfraind C and Vikkula M. Vascular Ependymoma. In: Atlas of Genetics and Cytogenetics in Oncology and Hematology 2009, 13(1):90-92. http://atlasgeneticsoncology.org//Genes/
Aerts A, Boon LM, Vikkula M. La génétique des anomalies vasculaires. Journal du Pédiatre Belge 2008, 10(2) :50-5.
Persu A & Vikkula M. Etudes d'association à l'echelle du génome: une révolution dans la génétique de l'athéromatose ? Athérosclérose Athérothrombose TransMed 2008, 49-60.
Vikkula, M. Identification des gènes et de leurs mutations responsables des anomalies vasculaires. L'agenda Cardio 2008, 21: 8-12.
Revencu N, Boon LM, Vikkula M. Arteriovenous malformation in mice and men. In : Tumor angiogenesis. Eds. D. Marmé & N. Fusenig, Springer Verlag, Germany (2008), 363-74.
Boon LM, Vikkula M. Vascular Anomalies. In : Fitzpatrick's Dermatology in General Medicine, 7th edition, McGraw-Hill Professional Publishing (2008), Editors: Klaus Wolff, Lowell A. Goldsmith, Stephen I. Katz, Barbara A. Gilchrest, Amy S. Paller, David J. Leffell. Vol 2: chapter 173: pp. 1651-66.
Wouters V, Boon LM, Vikkula M. TIE2 and cutaneomucosal venous malformation. In : Inborn errors of development. Ed. C.J. Epstein, R.P. Erickson, A. Wynshaw-Boris, Oxford University Press, New York, (2008), III, part E, pp. 491-4.
Revencu N, Boon LM, Mulliken JB, Vikkula M. RASA1 and capillary malformation-arteriovenous malformation. In : Inborn errors of development. Ed. C.J. Epstein, R.P. Erickson, A. Wynshaw-Boris, Oxford University Press, New York, (2008), III, part J, pp. 647-50.
Ghalamkarpour A, Devriendt K, Vikkula M. SOX18 and the hypotrichosis-lymphedema-telangiectasia syndrome. In : Inborn errors of development. Ed. C.J. Epstein, R.P. Erickson, A. Wynshaw-Boris, Oxford University Press, New York, (2008), IV, part E, pp. 913-5.
Brouillard P, Enjolras O, Boon LM, Vikkula M. Glomulin and glomuvenous malformation. In : Inborn errors of development. Ed. C.J. Epstein, R.P. Erickson, A. Wynshaw-Boris, Oxford University Press, New York, (2008), VI, pp. 1561-5.
Boon LM, Vikkula M. Multiple cutaneous and mucosal venous malformation. In: Pagon RA, Adam MP, Ardinger HH et al, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2015, 2008 Sep 18 [Updated 2015 Jan 29].
Vikkula M. Genetic causes of vascular anomalies. In: Journal des Maladies Vasculaires 2008, tome 33, hors-série n° 1.
Aerts V, Boon L, Vikkula M. Molecular genetics of hereditary vascular malformations. In: Encyclopedia of Life Sciences (ELS) (2009). John Wiley & Sons, Ltd, Chichester. DOI: 10.1002/9780470015902.a0021459.
Limaye N, Boon L, Vikkula M. From germline towards somatic mutations in the pathophysiology of vascular anomalies (review). Hum Mol Genet 2009, 18(R1) :R65-74. IF=7.39
Limaye N and Vikkula M. Genetic aspects of vascular malformations. In: Hemangiomas and Vascular Malformations - An Atlas of Diagnosis and Treatment, 1st edition. Editors: R. Mattassi, DA Loose, M. Vaghi (eds.), Springer-Verlag Italia 2009, Part III, chapter 13, pp 99-108.
Revencu N, Vikkula M. Cerebral cavernous malformation. In : Encyclopedia of molecular mechanisms of disease.

Editor: Lang F., Springer-Verlag Berlin GmbH, Heidelberg, Germany 2009.
Boon LM and Vikkula M. Blue jeans to blue genes. J Craniofac Surg 2009, 20(suppl 1) :703-6.
Persu A & Vikkula M. In: Athérosclérose Athérothrombose, 2010: 43-53. Ch 3: Les biomarqueurs de risque cardiovasculaire à l'heure de la génomique. Editors: H. Kulbertus, W. Van Mieghem and TransMed.
Gohy S, Pieters T, Goffette P, Vikkula M, Rodenstein D. Les manifestations vasculaires pulmonaires des télangiectasies hémorragiques héréditaires. Louvain médical 2010, 129:249-54.
Boon LM, Ballieux F, Vikkula M. Pathogenesis of vascular anomalies. Clin Plastic Surg 2011, 38(1):7-19. IF=1.422; 20 citations.
Boon LM, Enjolras O, Mulliken JB, Vikkula M. Vascular malformations. In: Harper, Textbook of Pediatric Dermatology, 3ed. Editors: Irvine A, Yan A and Hoeger P, Wiley-Blackwell 2011, vol 1, chapter 112, pp 112.1-112.24.
Storkebaum E, Quaegebeur A, Vikkula M and Carmeliet P. Cerebrovascular disorders: novel molecular insights and therapeutic opportunities (review). Nat Neurosci 2011, 14(11):1390-7. IF=15.351; 45 citations.
Uebelhoer M, Boon LM, Vikkula M. Vascular anomalies : from genetics towards models for therapeutic trials. In : Angiogenesis: From Development to Pathology. Editor: Michael Klagsbrun & Patricia A. D'Amore. Publisher: Cold Spring Harbor Laboratory Press, NY, USA, 2012, 2(8) :445-465.
Boon LM, Revencu N, Vikkula M. Capillary Malformation-Arteriovenous Malformation and RASA1 mutations. In : eLS. John Wiley & Sons, Ltd: Chichester. Published on line: 15 SEP 2011, DOI: 10.1002/9780470015902.a0022479
Boon LM, Vikkula M. Vascular Anomalies. In : Fitzpatrick's Dermatology in General Medicine, 8th edition, McGraw-Hill Professional Publishing. Editors: Klaus Wolff, Lowell A. Goldsmith, Stephen I. Katz, Barbara A. Gilchrest, Amy S. Paller, David J. Leffell. 2012, vol 2, section 28, chapter 172, p 2076-94.
Persu A and Vikkula M. Promesses et limites de la génétique cardiovasculaire: le locus 9p21.3. in: Kulbertus H and Van Mieghem W. Athérosclérose Athérothrombose TransMed 2013, 75-88.
Catala A, Roe E, Vikkula M, Baselga E. Capillary malformation-arteriovenous malformation syndrome: a report of 2 cases, diagnostic criteria and management. Actas Dermosifiliogr 2013, 104(8):710-3.
Brouillard P and Limaye N, Boon LM, Vikkula M. Disorders of the venous system. In: Emery and Rimoin's Principles and Practice in Medical Genetics, 6th edition, 2013. Chapter 56, 1-9.
Revencu N, Boon LM, Vikkula M. Capillary Malformation / Arteriovenous Malformation. In: Emery and Rimoin's Principles and Practice in Medical Genetics, 6th edition, 2013. Chapter 57, 1-7.
Boon LM and Vikkula M. Molecular genetics of vascular malformations. In: Mulliken & Young's Vascular Anomalies: Hemangiomas and Malformations, 2nd edition, Oxford University Press, New York, 2013, part 3, chap 9, pp 327-375. Editors: Mulliken JB, Burrows PE, Fishman S.
Bayet B, Revencu N, Castelein S, de Toeuf C, Reychler H, Bou Sabe SA, Theys S, Steenhout P, Bernard P, Vikkula M, François G. La fente labiopalatine et/ou palatine : signe d'appel d'un syndrome rare. Louvain médical 2013, 132:449-53.
Duhoux F, Vikkula M. La génomique en cancérologie expliquée à l'omnipraticien. Louvain médical 2014 ; 133(6) :417-21.
Brouillard P, Boon LM, Vikkula M. Genetics of lymphatic anomalies. J Clin Invest 2014, 124(3): 898-804. IF=13.215
Limaye N and Vikkula M. Molecular and genetic aspects of hemangiomas and vascular malformations. In: Hemangiomas and vascular malformations, 2nd edition. Editors: R. Mattassi, DA Loose and M. Vaghi, Springer Verlag, Milano, Italia, 2015, Part I, chapter 2, pp 21-38.
Ballieux F, Boon LM, Vikkula M. Blue Bleb Rubber Nevus syndrome: neurological implications. In: Handbook of Clinical Neurology 2015, 132:223-30.
Evenepoel L, Aydin S, Vikkula M, Persu A. La génétique des phéochromocytomes et paragangliomes: un domaine en pleine expansion. Institut Roi Albert II Newsletter 2015, 26:25-9. (vulgarisation)
Schlögel M, Brouillard P, Boon LM, Vikkula M. Genetic causes of lymphedema. In: Lymphedema: Presentation, Diagnosis and Treatment. A.K. Greene et al (eds), Springer International Publishing Switzerland 2015:19-31.
Dompmartin A, Revencu N, Boon LM, Vikkula M. Disorders affecting cutaneous vasculature. In: Griffiths et al, Rook's Textbook of Dermatology, 8th ed, section 5/chapter 73, 2016.
Nguyen H-L, Boon LM, Vikkula M. The genetic basis of vascular anomalies. In: Vascular Anomalies: A Guide for the Hematologist/Oncologist, Editors Drs. Trenor and Adams, Springer, in press.
Limaye N, Uebelhoer M, Boon LM, Mulliken JB, Vikkula M. TIE2 (TEK) and venous malformation. In : Inborn errors of development. Molecular basis of clinical disorders of morphogenesis. 3rd edition, Eds. R.P. Erickson, A. Wynshaw-Boris, Oxford University Press, New York. 2016, chapter IV: Other defined core developmental pathways, part C: The FGF signaling pathway, p. 493-6.
Revencu N, Boon LM, Mulliken JB, Vikkula M. RASA1 and capillary malformation-arteriovenous malformation. In: Inborn errors of development. Molecular basis of clinical disorders of morphogenesis. 3rd edition, Eds. R.P. Erickson, A. Wynshaw-Boris, Oxford University Press, New York. 2016, chapter IV: Other defined core developmental pathways, part H: The rasierkimapk pathway, p.633-8.
Brouillard P, Devriendt K, Vikkula M. SOX18 and the hypotrichosis-lymphedema-telangiectasia syndrome. In: Inborn errors of development. Molecular basis of clinical disorders of morphogenesis. 3rd edition, Eds. R.P. Erickson, A. Wynshaw-Boris, Oxford University Press, New York. 2016, chapter V: Transcription factors and chromatin regulators, part E: The SOX gene family, p. 867-70.
Brouillard P, Boon LM, Mulliken JB, Vikkula M. Glomulin and glomuvenous malformation. In : Inborn errors of development. Molecular basis of clinical disorders of morphogenesis. 3rd edition, Eds. R.P. Erickson, A. Wynshaw-Boris, Oxford University Press, New York. 2016, chapter VII: Dysmorphic disease genes of unkown function or unclassified, p.1423-8.
Schlögel M, Brouillard P, Boon LM, Vikkula M. Molecular genetics of lymphatic and complex vascular malformations. In : Lymphedema: A concise compendium of theory and practice. Section X: chapter 60. Eds: Byung-Boong Lee and Stanley G. Rockson, 2nd edition, Springer 2017.
Boon LM, Vikkula M & Seront E. Venous malformations. In : Moise L. Levy, UpToDate, 2017.
Boon LM & Vikkula M. Vascular malformations. In: A. Irvine, & P. Hoeger, A. Yan, Harper's Textbook of Pediatric Dermatology 4th edition. Wiley-Blackwell Publishing Ltd 2017.
Amyere M, Boon LM, Vikkula M. Genetics of arteriovenous malformations. In: Brain Arteriovenous Malformations: pathogenesis, epidemiology, diagnosis, treatment and outcome. Eds. Vladimir Benes & Ondrej Bradac. Springer 2017:23-36.
Nguyen H-L, Boon LM & Vikkula M. Genetics of vascular malformations. In: Mariella Catalano (IT), & Zsolt Pecsvarady (H), Dan Olinic (RO), Jean-Claude Wautrecht, Grigoris Gerotziafas, Beatrice Amann-Vesti, Debora Karetova, Bengt Fagrell, Curt Diehm, MatijaKozak, Michael Edmonds, VAS European Book on Angiology/Vascular Medicine 2017.
Queisser A, Boon LM, Vikkula M. Etiology and Genetics of Congenital Vascular Lesions. Otolaryngol Clin North Am 2018 Feb;51(1):41-53.
Boon LM, Ballieux F, Vikkula M. Vascular Malformations. In : Fitzpatrick's Dermatology in General Medicine, 9th edition, McGraw-Hill Education Publishing. Editors: Sewon Kang, Masayuki Amagai, Anna L. Bruckner, Alexander H. Enk, David J. Margolis, Amy J. McMichael, Jeffrey S. Orringer, 2019. Part 22, chapter 147: 2636-68.
Queisser A, Boon LM, Vikkula M. Molecular genetics of vascular malformations. In: eLS. John Wiley & Sons, Ltd: Chichester, 2018. DOI: 10.1002/9780470015902.a0021459.pub2
Boon LM & Vikkula M. Vascular malformations. In: A. Irvine, & P. Hoeger, A. Yan, Harper's Textbook of Pediatric Dermatology 5th edition. John Wiley & Sons, Ltd 2019, 1399-1424.
Brouillard P, Limaye N, Boon LM, Vikkula M. Disorders of the venous system. In: Emery and Rimoin's Principles and Practice in Medical Genetics, 7th edition, 2020, pp 251-260.
Revencu N, Boon LM, Vikkula M. Capillary Malformation / Arteriovenous Malformation. In: Emery and Rimoin's Principles and Practice in Medical Genetics, 7th edition, 2020, pp 261-266.
Dekeuleneer V, Seront E, Van Damme A, Boon LM, Vikkula M. Theranostic advances in vascular malformations (review). J Invest Dermatol 2020, 140:756-63.
Canaud G, Hammill AM, Adams D, Vikkula M, Keppler-Noreuil KM. A review of mechanisms of disease across PIK3CA-related disorders with vascular manifestations. Orphanet J Rare Dis. 2021 Jul 8;16(1):306.
Welsch S, Harvengta A, Gallo P, Martin M, Beckers D, Mouraux T, Seret N, Lebrethon MC, Helaers R, Brouillard P, Vikkula M, Lysy PA. A New Tool to Identify Pediatric Patients with Atypical Diabetes Associated with Gene

Polymorphisms. Diabetes Metab J. 2024 Mar 22. doi: 10.4093/dmj.2023.0166. Epub ahead of print. PMID: 38523249.
Dompmartin A, Revencu N, Boon LM, Vikkula M. Disorders affecting cutaneous vasculature. In: Griffiths et al, Rook's Textbook of Dermatology, 10th ed, part 6/chapter 71, 2024.
Seront E., Van Damme A, Coulie J, Dekeuleneer V, Vikkula M, Boon LM. Vascular malformations in 2023: Sirolimus confirms its efficacy and allows treatment in utero. Louvain Médical, 2024, 143:157-160.
Seront, E., Hermans, C., Boon, L. M., & Vikkula, M. (2024). Targeted treatments for vascular malformations: current state of the art. Journal of thrombosis and haemostasis : JTH, S1538-7836(24)00430-6. Advance online publication.
Coulie, J., Revencu, N., Boon, L.M., Vikkula, M. (2024). Capillary Malformation-Arteriovenous Malformation Syndrome. In: Rauen, K.A. (eds) The RASopathies. Springer, Cham.
Coulie J, Seront E, Vikkula M, Boon LM. Extracranial arteriovenous malformations: towards etiology-based therapeutic management. J Clin Invest. 2025 Mar 17;135(6):e172837.
Revencu N, Coulie J, Boon LM, Vikkula M. Genetic aspects of vascular malformations. Presse Med. 2025 Sep;54(3):104295.
Coulie J, Vikkula M, Boon LM. Peripheral arteriovenous malformations: Diagnosis and future prospects. Presse Med. 2025 Sep;54(3):104296.
Boutry S., Van Marcke C., Limaye N., Smits G., Brouillard P., De Bortoli M., Pairet E., Saadat Varnosfaderanii A., Helaers R., Fellay J., Lenaerts T., Vikkula M. DoAggregate: a pipeline for aggregation testing for rare-variants on a gene and pathway level. Abstracts from the 58th European Society of Human Genetics (ESHG) Conference: Hybrid Posters. Eur J Hum Genet 33 (Suppl 1), 508–1220 (2025).

Projects

Professor Miikka Vikkula, MD, PhD, is an internationally recognized leader in human and medical genetics, with a career spanning more than three decades at the interface of molecular genetics, vascular biology, and rare disease research. He received his medical and doctoral training at the University of Helsinki, completing an MD–PhD program in molecular genetics (MD 1992, PhD 1993), followed by licensure as a medical doctor in 1993. His early scientific training included international post-doctoral research fellowship in the United States, notably at Harvard Medical School (1993-1997), where he made seminal contributions to the genetic understanding of vascular anomalies. He later obtained the Agrégé de l'Enseignement Supérieur at the Université catholique de Louvain (UCLouvain, 2000) and was recognized as a specialist in clinical genetics by the Belgian Ministry of Health (2019).

Prof. Vikkula established his independent research group at the de Duve Institute in Brussels in 1997, where he has been a Full Member of the institute since 2004 and part of its Directorate since the same year. Academically, Prof. Vikkula has held successive professorial appointments at UCLouvain and has served as Full Professor of Human Genetics since 2013. He has also played major leadership roles, including Coordinator of the Centre for Human Genetics at Cliniques universitaires Saint-Luc (2011-2013). In parallel, he has contributed extensively to the scientific community through leadership in national and international societies (BeSHG, ESHG, NAVBO), peer review, and conference organization (BESHG, ESHG, NAVBO, ISSSVA, VAC), and through training the next generation of basic scientists and clinician-scientists.

His career has been distinguished by numerous prestigious honours and awards, including the INBEV–Baillet Latour Clinical Research Prize (2013), the first Generet Prize (2018), the Earl P. Benditt Award (NAVBO 2023), the LE&RN Lifetime Achievement Award (2024), the Gagna & Van Heck Prize for Incurable Diseases (2024), and the Black Pearl Scientific Award from EURORDIS (2025). He appeared on the Standford List of top 2% of scientists in 2021 and is ranked as a Highly Ranked Scholar among the top 0.05% of scholars worldwide in birth defects research by Scholar GPS. In recognition of his exceptional scientific and societal contributions, Prof. Vikkula was appointed Commander of the Order of Leopold by His Majesty King Philippe of Belgium in 2018.

Memberships

ISSVA, Vice President (10/05/2024)
LGDA (Lymphangiomatosis and Gorham's Disease Alliance) Medical and Clinical Advisory Council (27/10/2021-ongoing).
Cure-HHT Genetics Workstream (September 2021- April 2022)
PTEN foundation advisory board (2021 onwards)
ISSVA, Board Member at large (15/05/2020 – 10/05/2024)
NAVBO Meritorious Awards Committee (2015-2018)

Member of the Scientific Council of the International Society of Lymphology (ISL) (2023 -).
Member of the North American Vascular Biology Organisation (NAVBO) (2021-)
Full Member of the Académie Royale de Médecine de Belgique (04/07/2016 -).
Associate Member of the Académie Royale de Médecine de Belgique (24/11/2012 - 03/07/2016)
Member of the International Society for the Study of Vascular Anomalies (ISSVA) (1995 -).
Member of the American Society of Human Genetics (ASHG) (1997 -).
Member of the European Society of Human Genetics (ESHG) (1998 -).
Member of the Doctoral School of Biochemistry, Cellular Biology and Fundamental Microbiology, (BCM), Université catholique de Louvain, Brussels, Belgium (1999 -).
Member of the Doctoral School of Immunology and Genetics (GIM), Université catholique de Louvain, Brussels, Belgium (1999 -).
Member of the Belgian Society of Human Genetics (BeSHG) (2000 -).
Member of the American Heart Association Council on Basic Cardiovascular Research (2001 -).
Member of the Finnish Medical Association (Lääkäriliitto) (1987 -).
Member of the Finnish Medical Association Duodecim (Lääkäriseura Duodecim), Finlande (1987 - 2009).
Member of the Association of Young Physicians of Finland (1987 - 1999).
A PhD student member at "the Comity for Planning the M.D.-Ph.D. program", University of Helsinki, School of Medicine, 1993.

4. Honors and Awards

2025 The Black Pearl Scientific Award to recognise the major achievements and outstanding commitment of scientists who strive to make a difference for the rare disease community, EURORDIS-Rare diseases Europe (award ceremony 24/2/2025)

2024 Highly Ranked Scholar – Lifetime in the speciality of Birth Defects in recognition of exceptional productivity, noteworthy impact and quality of scholarly work in the top 0.05% of scholars in the Speciality worldwide. Conferred by Scholar GPS.

2024 The Gustav R. Born Honorary Award for Research in Angiology/Vascular Medicine, European Independent Foundation in Angiology / Vascular Medicine (with Prof. L. Boon)

2024 The triennial international Gagna and Van Heck Prize for incurable diseases, for their contribution to a better understanding of orphan vascular diseases, and for the significant therapeutic advances they have made (with Prof. L.M. Boon)

2024 Lifetime Achievement Award 2024 by the Lymphatic Education & Research Network (LE&RN) at Gordon Research Conference on Lymphatics

2023 – 2024 University of Louvain was ranked #1 in the ScholarGPS Academic Institution Rankings for 'Vascular Malformations' in the 'Prior 5 Years'

2023 The Earl P. Benditt Award, recognizing an individual who has made an outstanding discovery or developed a concept that has been seminal to our understanding of vascular biology or pathology, North American Vascular Biology Organisation (NAVBO), USA

2022 – 2026 European Co-ordinator of the Leducq Network of Excellence Grant: Recalibrating Mechanotransduction in Vascular Malformations

2019-2024 EU MSCA-ITN-2018, Co-ordinator of the VACure project grant

2011-2026 WELBIO, Walloon Excellence in Lifesciences and Biotechnology, Belgium

2018-2022 The First Generet Prize for the study of Rare Diseases (Fonds Roi Baudouin, Belgium)

2021 Appearance on the Standford List of top 2% of scientists by citation metrics

(doi: 10.17632/btchxktzyw.3)

2018 Awarded the rank of Commander of the Order of Leopold by His Majesty the King Philippe of Belgium.

2016 Prix du Concours ordinaire de la 1ère Section (Sciences Biomédicales), Académie Royale de Médecine, Belgium

2013 INBEV-BAILLET LATOUR Clinical Research Prize, Belgium (with Prof. L. Boon)

2010 The Lambertine-Lacroix Fundamental Sciences Prize in Cardiology for the characterization of the pathophysiological mechanisms underlying vascular anomalies, FNRS, Belgium

2006 Eugène de Sommer Scientific Award, University catholique de Louvain, Belgium

2006 Pfizer Scientific Award, Belgium

Nov 2004 11th Manchester Birth Defects Conference, Manchester, UK. Keynote lecture: "Genetic causes of vascular malformations »

Feb 2004 15th International Workshop on Vascular Anomalies, Wellington, New Zealand. Keynote Lecture: "Molecular Basis of Vascular Anomalies".

June 2003 The Society for Pediatric Dermatology Annual Meeting, Seattle, USA. Sidney Hurwitz keynote lecture: "Molecular genetics of vascular anomalies".

June 2002 14th International Workshop on Vascular Anomalies, Nijmegen, Holland. Keynote Lecture: "Towards understanding the ethiopathogenesis of vascular malformations".

2002 Pharmacia Scientific Award, Belgium

1998-2000 Marie-Curie fellowship award of the European Union

1997 HFSPO Long-Term Fellowship Award (2-years)

1994-1996 Fogarty International Fellowship Award, NIH, USA

Invited congress presentations (total >300)

2025 Invited congress presentations:

7/3/2025, Filière Santé Maladies Rares TeteCou, Journée Recherche et Innovation, Paris, France, Invited presentation: Vascular malformations: from genetic causes, to preclinical models and clinical trials
7-11/4/2025, 15th World Congress of Pediatric Dermatology (WCPD), Buenos Aeres, Argentina, Invited Presentation (with Prof. L. Boon): Genetic Basis and Therapies for Vascular Anomalies
25/4/2025, European Lymphatic Symposium (ELS), Toulouse, France, Invited presentation: Novel insights into the pathophysiology of lymphedema
26/4/2025, Wroclaw Polish Lymphatic meeting, Wroclaw, Poland, Invited presentation: Genetics aspects of primary lymphedema and lymphatic malformations
9/6/2025, WRI symposium, Helsinki, Finland: Lymphatic anomalies: From genes and pathophysiology towards therapeutic trials.

12-14/6/2025, Lymphatic Forum 2025, Chicago, USA: Lymphatic anomalies: From genes and pathophysiology towards therapeutic trials
10-12/09/2025, BeNeVBO meeting, Leuven, Belgium: Vascular malformations: from genetic causes, to preclinical models and clinical trials
14-16/09/2025, DEF 2025, Paris, France: Genetics of polymalformative syndromes involving the orbit and nose
19/09/2025, 4th Annual Lymphatic conference, Children's Hospital of Philadelphia, USA: Beyond the Periphery: Searching for Clues for Central Conduction Defects in Lymphedema
3/10/2025, Annual Czech-Slovak pediatric hematology oncology conference, Brno, Czech Republic: VASCERN, the European Reference Network (ERN) for Rare Multisystemic Vascular Diseases VASCA (Vascular Anomaly) Working Group
15-19/10/2025, 30th ISL World Congress of Lymphology, Antalya, Türkiye: "Sex-specific penetrance of pathogenic variants in primary lymphedema" and "Novel genetic findings on heterogeneity of primary lymphedema" Best oral presentation award
27-29/10/2025, World Orphan Drug Congress, Amsterdam, The Netherlands: From Malignancy to Malformation: Navigating the Hurdles of Cancer Drug Repositioning for Vascular Anomalies.
02-03/11/2025, European Course in Neuroradiology, Valencia, Spain: Vessels, genes and pathways: are we targeting the correct physiopathology
07-10/11/2025, AHA Scientific Sessions, New Orleans, United States: Human Lymphedema: Expanding Complex Genetics and Disrupting Pathophysiological Pathways
30/11-03/12/2025, BRAIN conference, London, United Kingdom: Genetics of AVMs
04-06/12/2025, Congrès de la société française de lymphologie, Toulouse, France: Lymphatic malformations
03-04/02/2026, Académie Nationale de Médecine, Paris, France : Données récentes sur la pathophysiologie des malformations vasculaires périphériques

Other Relevant Information

Prof. Vikkula is internationally recognized as a leading authority on the molecular basis of vascular anomalies and a pioneer in the development of targeted therapies. He has published more than 250 peer-reviewed original research

articles and over 80 reviews and book chapters in major medical textbooks. His work has been cited over 31,000 times, resulting in an H-index of 89 (Google Scholar).

Prof. Vikkula's research group has developed and applied state-of-the-art genomics and transcriptomics approaches, including high-throughput next-generation sequencing (NGS), in numerous national and international collaborative projects. The group has also established a major vascular anomalies tissue biobank, which has become a key resource for molecular and pathological characterization of these disorders. In parallel, extensive in vivo studies using genetically engineered mouse models have been conducted to investigate disease mechanisms and therapeutic strategies.

His work has led to the discovery of the genetic defects underlying multiple vascular anomalies and has substantially advanced the field, enabling revision of disease classification (Wassef et al., 2015) and the identification of novel clinical entities (Eerola et al., 2003; Amyere et al., 2017). Using high-throughput NGS, his team identified several novel genes implicated in vascular anomalies, including EPHB4 (Amyere et al., 2017) and PIK3CA (Limaye et al., 2015). They also expanded the phenotypic spectrum associated with mutations in known genes, such as TIE2, which they demonstrated to be mutated in a large proportion of sporadic venous malformations, as well as in blue rubber bleb nevus syndrome and multifocal sporadic venous malformations (Soblet et al., 2017).

Notably, Prof. Vikkula's group generated the first mouse model of venous malformation (Boscolo et al., 2015) and demonstrated therapeutic efficacy using the mTOR inhibitor rapamycin. These findings directly led to phase 2A (Boscolo et al., 2015) and phase 2B (Hammer et al., 2018) clinical trials. A phase 3 trial is currently ongoing, with approximately 100 patients enrolled to date (VASE, EudraCT 2015-001703-32).

Supported by highly competitive national and international funding, including FNRS, EU Marie Curie, Leducq Network of Excellence, MSCA-ITN, and WELBIO programs, Prof. Vikkula has built a world-leading research group that pioneered the discovery of germline and somatic mutations underlying vascular malformations and elucidated key pathophysiological mechanisms, including second-hit events and mechanotransduction. His work has had a transformative impact on the diagnosis and treatment of rare vascular diseases.