Anne JOUTEL

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Education and Qualifications

Medicine

- Board certified in Medicine, Faculty of Medicine Rouen (1988)
- Board certified in Neurology, University Paris Diderot-Paris 7 (1994)
- M.D., University Paris Diderot-Paris 7 (1996)

Science

- M.S. (Neurosciences/Genetics), University Pierre et Marie Curie-Paris 6 (1992)
- Ph.D. (Neurosciences/Genetics), University Pierre et Marie Curie-Paris 6 (1996)
- HDR "Habilitation à diriger des recherches", University Paris Diderot-Paris 7 (2001)

Positions and Employment

- Residency in Neurology, Assistance Publique des Hôpitaux de Paris (1988-1993)
- Research fellowship, Paris (1993-1996)
- Post-doctoral Research fellowship, Paris (1996-1998)
- INSERM Junior Researcher (CR1) (1998-2005)
- Consultant, Dept of Genetics, Lariboisière Hospital, Paris (2002-2011)
- INSERM Research Director (DR2) (2006-present)
- Group leader "Pathogenesis of small vessel diseases of the brain" (2012-present)

Honors and Awards

- Internat des Hôpitaux de Paris (rank 15) (1988)
- Award from the French Neurological Society (1993)
- INSERM-Assistance Publique des Hôpitaux de Paris Research fellowship (1993-1996)
- Clara and Victor Soriano prize from the National Academy of Medicine (1996)
- Hélène Anavi prize from Fondation JEAN DAUSSET (1997)
- JEAN VALADE Young investigator prize from Fondation de France (2000)
- Anita Harding prize from the European Society of Neurology (2001)
- INSERM- Assistance Publique des Hôpitaux de Paris award for translational research (2002-2011)
- RO1 NIH grant from USA (2006-2010)

Honorary activities

- Member of the INSERM Scientific Committee "CSS4, Pathophysiology " (2008-2012)
- Member of the Evaluation Committee « Pathophysiology » at Agence Nationale de la Recherche (2014 -2015)
- Member of the Scientific Committee of CADASIL France

Publications

H-index: 43; 7996 citations (web of science)

Peer reviewed papers

1. **Anne Joutel**, Hugues Chabriat. Pathogenesis of white matter changes in cerebral small vessel diseases: beyond vessel-intrinsic mechanisms. Clin Sci (Lond). 2017 Apr 25;131(8):635-651. doi: 10.1042/CS20160380. Review
2. Baron-Menguy C, Domenga-Denier V, Ghezali L, Faraci FM, **Joutel A**. Increased Notch3 Activity Mediates Pathological Changes in Structure of Cerebral Arteries. **Hypertension. 2017** Jan;69(1):60-70.

3. Capone C, Dabertrand F, Baron-Menguy C, Chalaris A, Ghezali L, Domenga-Denier V, Schmidt S, Huneau C, Rose-John S, Nelson MT, **Joutel A.** Mechanistic insights into a TIMP3-sensitive pathway constitutively engaged in the regulation of cerebral hemodynamics. **Elife. 2016** Aug 1;5. pii: e17536. doi: 10.7554/eLife.17536.
4. Capone C, Cognat E, Ghezali L, Baron-Menguy C, Aubin D, Mesnard L, Stöhr H, Domenga-Denier V, Nelson MT, **Joutel A**. Reducing Timp3 or vitronectin ameliorates disease manifestations in CADASIL mice. **Ann Neurol., 2016** Mar;79(3):387-403.
5. Rosenberg GA, Wallin A, Wardlaw JM, Markus HS, Montaner J, Wolfson L, Iadecola C, Zlokovic BV, **Joutel A**, Dichgans M, Duering M, Schmidt R, Korczyn AD, Grinberg LT, Chui HC, Hachinski V.Consensus statement for diagnosis of subcortical small vessel disease**. J Cereb Blood Flow Metab. 2016** Jan;36(1):6-25.
6. **Joutel A**, Haddad I, Ratelade J, Nelson MT. Perturbations of the cerebrovascular matrisome: a convergent mechanism in small vessel disease of the brain? **J Cereb Blood Flow Metab. 2016** Jan;36(1):143-57..
7. Dabertrand F, Krøigaard C, Bonev AD, Cognat E, Dalsgaard T, Domenga-Denier V, Hill-Eubanks DC, Brayden JE, **Joutel A**, Nelson MT. Potassium channelopathy-like defect underlies early-stage cerebrovascular dysfunction in a genetic model of small vessel disease. **Proc Natl Acad Sci U S A**. **2015** Feb 17;112(7):E796-805. doi: 10.1073/pnas.1420765112. Epub 2015 Feb 2.
8. Tikka S, Baumann M, Siitonen M, Pasanen P, Pöyhönen M, Myllykangas L, Viitanen M, Fukutake T, Cognat E, **Joutel A***, Kalimo H*. CADASIL and CARASIL. **Brain Pathol. 2014** Sep;24(5):525-44. doi: 10.1111/bpa.12181. (* Shared senior authorship)
9. Kast J, Hanecker P, Beaufort N, Giese A, **Joutel A**, Dichgans M, Opherk C, Haffner C. Sequestration of latent TGF-β binding protein 1 into CADASIL-related Notch3-ECD deposits. **Acta Neuropathol Commun. 2014** Aug 13;2(1):96. [Epub ahead of print]
10. Cognat E, Hervé D, **Joutel A**. Response to Letter Regarding Article, "Archetypal Arg169Cys Mutation in NOTCH3 Does Not Drive the Pathogenesis in Cerebral Autosomal Dominant Arteriopathy With Subcortical Infarcts and Leucoencephalopathy via a Loss-of-Function Mechanism" **Stroke**. **2014** Jul;45(7):e129. doi: 10.1161/STROKEAHA.114.005616. Epub 2014 May 27.
11. Cognat E, Cleophax S, Domenga-Denier V, **Joutel A**. Early white matter changes in CADASIL: evidence of segmental intramyelinic oedema in a pre-clinical mouse model **Acta Neuropathol Commun. 2014**, **2**:49 (epub)
12. **Joutel A**, Faraci FM. Cerebral small vessel disease (SVD): Insights and opportunities from mouse models of collagen IV-related SVD and CADASIL. **Stroke 2014** Apr;45(4):1215-21.
13. Cognat E, Baron-Menguy C, Domenga-Denier V, Cleophax S, Fouillade C, Monet-Leprêtre M, Dewerchin M, **Joutel A**. The archetypal Arg169Cys mutation in NOTCH3 does not drive the pathogenesis in CADASIL via a loss of function mechanism. **Stroke 2014** Mar;45(3):842-9.
14. Dichgans M, **Joutel A**, Chabriat H. Letter by Dichgans et al regarding article, "Peripheral artery disease as a manifestation of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) and practical implications". **Circulation. 2013** Oct 22;128(17):e363.
15. **Joutel A** Loss-of-function mutation in the NOTCH3 gene: simply a polymorphism? **Hum Mutat. 2013** Nov;34(11):v. doi: 10.1002/humu.22198.
16. Monet-Leprêtre M, Haddad I, Baron-Menguy C, Fouillot-Panchal M, Riani M, Domenga-Denier V, Dussaule C, Cognat E, Vinh J, **Joutel A**. Abnormal recruitment of extracellular matrix proteins by excess Notch3ECD: a new pathomechanism in CADASIL. **Brain. 2013** Jun;136(Pt 6):1830-45.
17. Fouillade C, Baron-Menguy C, Domenga-Denier V, Thibault C, Takamiya K, Huganir R, **Joutel A**. Transcriptome Analysis of Notch3 Target Genes Identifies Grip2 as a Novel Regulator of Myogenic Response in the Cerebrovasculature. **Arterioscler Thromb Vasc Biol. 2013** Jan;33(1):76-86.
18. Fouillade C, Monet-Leprêtre M, Baron-Menguy C & **Joutel A**. Notch signaling in smooth muscle cells during development and disease. **Cardiovasc Res. 2012** Jul 15;95(2):138-46.
19. Caplan LR, Arenillas J, Cramer SC, **Joutel A**, Lo EH, Meschia J, Savitz S, Tournier-Lasserve E. Stroke-Related Translational Research. **Arch Neurol. 2011** Sep;68(9):1110-23.

20. Boulos N, Helle F, Dussaule JC, Placier S, Milliez P, Djudjaj S, Guerrot D, **Joutel A**, Ronco P, Boffa JJ, Chatziantoniou C. Notch3 is essential for regulation of the renal vascular tone. **Hypertension**. **2011** Jun;57(6):1176-82.
21. Eikermann-Haerter K, Yuzawa I, Dilekoz E, **Joutel A,** Moskowitz MA and Ayata C. CADASIL mutations increase suceptibility to spreading depression. **Ann Neurol. 2011** Feb;69(2):413-8.
22. Joutel A and Debette S. Mendelian and complex causes of migraine: bridging the gap. **Cephalalgia**
23. **Joutel A**. Pathogenesis of CADASIL : Transgenic and knock-out mice to probe function and dysfunction of the mutated gene, Notch3, in the cerebrovasculature. **Bioessays**. **2011** Jan;33(1):73-80
24. Joutel A, Monet-Leprêtre M, Gosele C, Baron-Menguy, Hammes A, Schmidt S, Lemaire-Carrette, Domenga V, Schedl A, Lacombe P & Hubner N. Cerebrovascular dysfunction and microcirculation rarefaction precede white matter lesions in a mouse model of CADASIL. **J Clin Invest 2010** Feb 1;120(2):433-45
25. Chabriat H*, Joutel A*, Dichgans M*, Tournier-Lasserve E, Bousser MG. CADASIL. **Lancet Neurology 2009** Jul;8(7):643-53 (* Shared first authorship)
26. Monet-Leprêtre M, Bardot B, Lemaire B, Domenga V, Godin O, Dichgans M, Tournier-Lasserve E, Cohen-Tannoudji M, Chabriat H., Joutel A. Distinct phenotypic and functional features of CADASIL mutations in the Notch3 ligand binding domain. **Brain 2009**; Jun;132(Pt 6):1601-12
27. Tikka S, Mykkänen K, Ruchoux MM, Bergholm R, Junna M, Pöyhönen M, Yki-Järvinen H, Joutel A, Viitanen M, Baumann M, Kalimo H. Congruence between NOTCH3 mutations and GOM in 131 CADASIL patients. B**rain. 2009** Apr;132(Pt 4):933-9
28. Belin de Chantemèle EJ, Retailleau K., Pinaud F., Vessières E., Bocquet A, Guihot A.L., Lemaire B., Domenga V., Baufreton C., Loufrani L., Joutel A., Henrion D. Notch3 is a major regulator of vascular tone in cerebral and tail resistance arteries. **Arterioscler Thromb Vasc Biol. 2008** Dec;28(12):2216-2224.
29. Fouillade C, Chabriat H, Riant F, Mine M, Arnoud M, Magy L, Bousser MG, Tournier-Laserve E, Joutel A. Activating Notch3 mutation in a patient with samll-vessel-disease of the brain. **Hum Mutat. 2008** Mar;29(3):452 [Epub ahead of print]
30. Monet M, Domenga V, Lemaire B, Souilhol C, Langa F, Babinet C, Gridley T, Tournier-Lasserve E, Cohen-Tannoudji M, Joutel A.The archetypal R90C CADASIL-NOTCH3 mutation retains NOTCH3 function in vivo. **Hum Mol Genet. 2007** Apr 15;16(8):982-92.
31. Ishida C, Sakajiri K, Yoshita M, Joutel A, Cave-Riant F, Yamada M. CADASIL with a novel mutation in exon 7 of NOTCH3 (C388Y). **Intern Med. 2006**;45(16):981-5.
32. Souilhol C, Cormier S, Monet M, Vandormael-Pournin S, Joutel A, Babinet C, Cohen-Tannoudji M.Nas transgenic mouse line allows visualization of Notch pathway activity in vivo. **Genesis. 2006** Jun;44(6):277-86.
33. Verreault S, Joutel A, Riant F, Neves G, Rui Silva M, Maciazek J, Tournier-Lasserve E, Bousser MG, Chabriat H. A novel hereditary small vessel disease of the brain. **Ann Neurol. 2006** Feb;59(2):353-7.
34. Lacombe P, Oligo C, Domenga V, Tournier-Lasserve E, Joutel A. Impaired Cerebral Vasoreactivity in a Transgenic Mouse Model of Cerebral Autosomal Dominant Arteriopathy With Subcortical Infarcts and Leukoencephalopathy Arteriopathy. **Stroke. 2005** May;36(5):1053-8.
35. Dubroca C, Lacombe P, Domenga V, Maciazek J, Levy B, Tournier-Lasserve E, Joutel A, Henrion D. Impaired vascular mechanotransduction in a transgenic mouse model of CADASIL arteriopathy. **Stroke. 2005** *Jan;36(1):113-7.*
36. Domenga V, Fardoux P, Lacombe P, Monet M, Maciazek J, Krebs LT, Klonjkowski B, Berrou E, Mericskay M, Li Z, Tournier-Lasserve E, Gridley T, Joutel A. Notch3 is required for arterial identity and maturation of vascular smooth muscle cells. **Genes Dev. 2004** *Nov 15;18(22):2730-5.*
37. Vahedi K, Chabriat H, Levy C, **Joutel A**, Tournier-Lasserve E, Bousser MG. Migraine with aura and brain magnetic resonance imaging abnormalities in patients with CADASIL. **Arch Neurol. 2004** *Aug;61(8):1237-40.*
38. Denier C, Labauge P, Brunereau L, Cave-Riant F, Marchelli F, Arnoult M, Cecillon M, Maciazek J, **Joutel A**, Tournier-Lasserve E; Societe Francaise de Neurochirgurgie; Societe de Neurochirurgie de

Langue Francaise. Clinical features of cerebral cavernous malformations patients with KRIT1 mutations. **Ann Neurol. 2004** *Feb;55(2):213-20.*

39. **Joutel A**, Monet M, Domenga V, Riant F, Tournier-Lasserve E. Pathogenic mutations associated with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy differently affect Jagged1 binding and Notch3 activity via the RBP/JK signaling Pathway. **Am J Hum Genet. 2004** *Feb;74(2):338-47.*
40. Krebs LT, Xue Y, Norton CR, Sundberg JP, Beatus P, Lendahl U, **Joutel A**, Gridley T. Characterization of Notch3-deficient mice: normal embryonic development and absence of genetic interactions with a Notch1 mutation. **Genesis. 2003** Nov;37(3):139-43.
41. Vahedi K, Massin P, Guichard JP, Miocque S, Polivka M, Goutieres F, Dress D, Chapon F, Ruchoux MM, Riant F, **Joutel A**, Gaudric A, Bousser MG, Tournier-Lasserve E. Hereditary infantile hemiparesis, retinal arteriolar tortuosity, and leukoencephalopathy. **Neurology. 2003** Jan 14;60(1):57-63.
42. Ruchoux MM, Domenga V, Brulin P, Maciazek J, Limol S, Tournier-Lasserve E, **Joutel A**. Transgenic mice expressing mutant Notch3 develop vascular alterations characteristic of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. **Am J Pathol. 2003** Jan;162(1):329-42.
43. Cave-Riant F, Denier C, Labauge P, Cecillon M, Maciazek J, **Joutel A**, Laberge-Le Couteulx S, Tournier-Lasserve E. Spectrum and expression analysis of KRIT1 mutations in 121 consecutive and unrelated patients with Cerebral Cavernous Malformations. **Eur J Hum Genet. 2002** Nov;10(11):733-40.
44. Denier C, Gasc JM, Chapon F, Domenga V, Lescoat C, **Joutel A**, Tournier-Lasserve E. Krit1/cerebral cavernous malformation 1 mRNA is preferentially expressed in neurons and epithelial cells in embryo and adult. **Mech Dev. 2002** Sep;117(1-2):363-7.
45. **Joutel A**, Favrole P, Labauge P, Chabriat H, Lescoat C, Andreux F, Domenga V, Cecillon M, Vahedi K, Ducros A, Cave-Riant F, Bousser MG, Tournier-Lasserve E. Skin biopsy immunostaining with a Notch3 monoclonal antibody for CADASIL diagnosis. **Lancet. 2001** Dec 15;358(9298):2049-51.
46. Ducros A, Denier C, **Joutel A**, Cecillon M, Lescoat C, Vahedi K, Darcel F, Vicaut E, Bousser MG, Tournier-Lasserve E. The clinical spectrum of familial hemiplegic migraine associated with mutations in a neuronal calcium channel. **N Engl J Med. 2001** Jul 5;345(1):17-24.
47. **Joutel A**, Chabriat H, Vahedi K, Domenga V, Vayssiere C, Ruchoux MM, Lucas C, Leys D, Bousser MG, Tournier-Lasserve E. Splice site mutation causing a seven amino acid Notch3 in-frame deletion in CADASIL. **Neurology. 2000** May 9;54(9):1874-5.
48. **Joutel A**, Dodick DD, Parisi JE, Cecillon M, Tournier-Lasserve E, Bousser MG. De novo mutation in the Notch3 gene causing CADASIL. **Ann Neurol. 2000** Mar;47(3):388-91.
49. **Joutel A**, Andreux F, Gaulis S, Domenga V, Cecillon M, Battail N, Piga N, Chapon F, Godfrain C, Tournier-Lasserve E. The ectodomain of the Notch3 receptor accumulates within the cerebrovasculature of CADASIL patients. **J Clin Invest. 2000** Mar;105(5):597-605.
50. Heinzlef O, Alamowitch S, Sazdovitch V, Chillet P, **Joutel A**, Tournier-Lasserve E, Roullet E. Autoimmune diseases in families of French patients with multiple sclerosis. **Acta Neurol Scand. 2000** Jan;101(1):36-40.
51. Escary JL, Cecillon M, Maciazek J, Lathrop M, Tournier-Lasserve E, **Joutel A**. Evaluation of DHPLC analysis in mutational scanning of Notch3, a gene with a high G-C content. **Hum Mutat. 2000** Dec;16(6):518-26.
52. Echenne B, Ducros A, Rivier F, **Joutel A**, Humbertclaude V, Roubertie A, Azais M, Bousser MG, Tournier-Lasserve E. Recurrent episodes of coma: an unusual phenotype of familial hemiplegic migraine with linkage to chromosome 1. **Neuropediatrics. 1999** Aug;30(4):214-7.
53. Laberge-le Couteulx S, Jung HH, Labauge P, Houtteville JP, Lescoat C, Cecillon M, Marechal E, **Joutel A**, Bach JF, Tournier-Lasserve E. Truncating mutations in CCM1, encoding KRIT1, cause hereditary cavernous angiomas. **Nat Genet. 1999** Oct;23(2):189-93.

54. Denier C, Ducros A, Vahedi K, **Joutel A**, Thierry P, Ritz A, Castelnovo G, Deonna T, Gerard P, Devoize JL, Gayou A, Perrouty B, Soisson T, Autret A, Warter JM, Vighetto A, Van Bogaert P, Alamowitch S, Roullet E, Tournier-Lasserve E. High prevalence of CACNA1A truncations and broader clinical spectrum in episodic ataxia type 2. **Neurology. 1999** Jun 10;52(9):1816-21.
55. Chabriat H, Mrissa R, Levy C, Vahedi K, Taillia H, Iba-Zizen MT, **Joutel A**, Tournier-Lasserve E, Bousser MG. Brain stem MRI signal abnormalities in CADASIL. **Stroke. 1999** Feb;30(2):457-9.
56. Ducros A, Denier C, **Joutel A**, Vahedi K, Michel A, Darcel F, Madigand M, Guerouaou D, Tison F, Julien J, Hirsch E, Chedru F, Bisgard C, Lucotte G, Despres P, Billard C, Barthez MA, Ponsot G, Bousser MG, Tournier-Lasserve E. Recurrence of the T666M calcium channel CACNA1A gene mutation in familial hemiplegic migraine with progressive cerebellar ataxia. **Am J Hum Genet. 1999** Jan;64(1):89-98.
57. **Joutel A**, Tournier-Lasserve E. Notch signalling pathway and human diseases. **Semin Cell Dev Biol. 1998** Dec;9(6):619-25.
58. Furby A, Vahedi K, Force M, Larrouy S, Ruchoux MM, **Joutel A**, Tournier-Lasserve E. Differential diagnosis of a vascular leukoencephalopathy within a CADASIL family: use of skin biopsy electron microscopy study and direct genotypic screening**. J Neurol. 1998** Nov;245(11):734-40.
59. Chabriat H, Levy C, Taillia H, Iba-Zizen MT, Vahedi K, **Joutel A**, Tournier-Lasserve E, Bousser MG. Patterns of MRI lesions in CADASIL. Neurology. 1998 Aug;51(2):452-7. Djouhri H, Marsot-Dupuch K, Joutel A, Kujas M, Brette MD, Artuis F, Tubiana JM. Perichiasmatic granuloma occuring after radical mastoidectomy: MR findings. **Eur Radiol. 1998**;8(2):286-8.
60. Ducros A, **Joutel A**, Vahedi K, Cecillon M, Ferreira A, Bernard E, Verier A, Echenne B, Lopez de Munain A, Bousser MG, Tournier-Lasserve E. Mapping of a second locus for familial hemiplegic migraine to 1q21-q23 and evidence of further heterogeneity. **Ann Neurol. 1997** Dec;42(6):885-90.
61. **Joutel A**, Vahedi K, Corpechot C, Troesch A, Chabriat H, Vayssiere C, Cruaud C, Maciazek J, Weissenbach J, Bousser MG, Bach JF, Tournier-Lasserve E. Strong clustering and stereotyped nature of Notch3 mutations in CADASIL patients. **Lancet. 1997** Nov 22;350(9090):1511-5.
62. **Joutel A**, Ducros A, Alamowitch S, Cruaud C, Domenga V, Marechal E, Vahedi K, Chabriat H, Bousser MG, Tournier-Lasserve E. A human homolog of bacterial acetolactate synthase genes maps within the CADASIL critical region. **Genomics. 1996** Dec 1;38(2):192-8.
63. **Joutel A**, Corpechot C, Ducros A, Vahedi K, Chabriat H, Mouton P, Alamowitch S, Domenga V, Cecillion M, Marechal E, Maciazek J, Vayssiere C, Cruaud C, Cabanis EA, Ruchoux MM, Weissenbach J, Bach JF, Bousser MG, Tournier-Lasserve E. Notch3 mutations in CADASIL, a hereditary adult-onset condition causing stroke and dementia. **Nature. 1996** Oct 24;383(6602):707-10.
64. Ducros A, Nagy T, Alamowitch S, Nibbio A, **Joutel A**, Vahedi K, Chabriat H, Iba-Zizen MT, Julien J, Davous P, Goas JY, Lyon-Caen O, Dubois B, Ducrocq X, Salsa F, Ragno M, Burkhard P, Bassetti C, Hutchinson M, Verin M, Viader F, Chapon F, Levasseur M, Mas JL, Delrieu O, et al. Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy, genetic homogeneity, and mapping of the locus within a 2-cM interval. **Am J Hum Genet. 1996** Jan;58(1):171-81.
65. Chabriat H, Vahedi K, Iba-Zizen MT, **Joutel A**, Nibbio A, Nagy TG, Krebs MO, Julien J, Dubois B, Ducrocq X, et al. Clinical spectrum of CADASIL: a study of 7 families. Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. **Lancet. 1995** Oct 7;346(8980):934-9.
66. Ducros A, **Joutel A**, Labauge P, Pages M, Bousser MG, Tournier-Lasserve E. Monozygotic twins discordant for familial hemiplegic migraine. **Neurology. 1995** Jun;45(6):1222.
67. Chabriat H, Tournier-Lasserve E, Vahedi K, Leys D, **Joutel A**, Nibbio A, Escaillas JP, Iba-Zizen MT, Bracard S, Tehindrazanarivelo A, et al. Autosomal dominant migraine with MRI white-matter abnormalities mapping to the CADASIL locus. **Neurology. 1995** Jun;45(6):1086-91.
68. Vahedi K, **Joutel A**, Van Bogaert P, Ducros A, Maciazeck J, Bach JF, Bousser MG, Tournier-Lasserve E. A gene for hereditary paroxysmal cerebellar ataxia maps to chromosome 19p. **Ann Neurol. 1995** Mar;37(3):289-93.

69. **Joutel A**, Ducros A, Vahedi K, Labauge P, Delrieu O, Pinsard N, Mancini J, Ponsot G, Gouttiere F, Gastaut JL, et al. Genetic heterogeneity of familial hemiplegic migraine. **Am J Hum Genet. 1994** Dec;55(6):1166-72.
70. **Joutel A**, Bousser MG, Biousse V, Labauge P, Chabriat H, Nibbio A, Maciazek J, Meyer B, Bach MA, Weissenbach J, et al. A gene for familial hemiplegic migraine maps to chromosome 19. **Nat Genet. 1993** Sep;5(1):40-5.
71. Tournier-Lasserve E, **Joutel A**, Melki J, Weissenbach J, Lathrop GM, Chabriat H, Mas JL, Cabanis EA, Baudrimont M, Maciazek J, et al. Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy maps to chromosome 19q12. **Nat Genet. 1993** Mar;3(3):256-9.
72. Baudrimont M, Dubas F, **Joutel A**, Tournier-Lasserve E, Bousser MG. Autosomal dominant leukoencephalopathy and subcortical ischemic stroke. A clinicopathological study. **Stroke. 1993** Jan;24(1):122-5.
73. **Joutel A**, Moulonguet A, Demaugre F, Janowski M, Lacroix-Jousselin C, Said G. [Type II carnitine palmitoyl transferase deficiency complicated by acute respiratory failure] **Rev Neurol (Paris). 1993**;149(12):797-9.

Didactic publications

1. **Joutel A**, Tournier-Lasserve E. [Molecular basis and physiopathogenic mechanisms of CADASIL: a model of small vessel diseases of the brain] **J Soc Biol. 2002**;196(1):109-15. Review.
2. **Joutel A**, Francois A, Chabriat H, Vahedi K, Andreux F, Domenga V, Cecillon M, Maciazek J, Bousser MG, Tournier-Lasserve E. [CADASIL: genetics and physiopathology] **Bull Acad Natl Med. 2000**;184(7):1535-42; discussion 1542-4. Review.
3. Chabriat H, **Joutel A**, Vahedi K, Iba-Zizen MT, Tournier-Lasserve E, Bousser MG. [CADASIL (cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy): clinical features and neuroimaging] **Bull Acad Natl Med. 2000**;184(7):1523-31; discussion 1531-3. Review.
4. Chabriat H, **Joutel A**, Vahedi K, Iba-Zizen MT, Tournier-Lasserve E, Bousser MG. [CADASIL. Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalophathy] **Rev Neurol (Paris). 1997** Jul;153(6-7):376-85. Review.
5. Chabriat H, **Joutel A**, Vahedi K, Iba-Zizen MT, Tournier-Lasserve E, Bousser MG. [CADASIL (cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy)] **J Mal Vasc. 1996**;21(5):277-82. Review.
6. **Joutel A**, Tournier-Lasserve E, Bousser MG. [Hemiplegic migraine] **Presse Med. 1995** Feb 25;24(8):411-4. Review
7. **Joutel A**, Bousser MG, Biousse V, Labauge P, Chabriat H, Nibbio A, Maciazek J, Meyer B, Bach MA, Weissenbach J, et al. [Familial hemiplegic migraine. Localization of a responsible gene on chromosome 19] **Rev Neurol (Paris). 1994**;150(5):340-5.

Book chapters

1. H. Chabriat, **A. Joutel**, K. Vahedi, E. Tournier-Lasserve & MGB Bousser "CADASIL, Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy" in STROKE Pathophysiology, Diagnosis, and Management, Third Edition (Churchill Livingstone)
2. M. Dichgans & **A. Joutel** "Cerebrovascular Disorders" in Emery and Rimoin's principles and practice of medical genetics, fourth edition volIII, 3209-3230
3. **Anne Joutel** (2009). CADASIL (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy). In: Squire LR (ed.) Encyclopedia of Neuroscience, volume 2, pp. 481-487. Oxford: Academic Press
4. **Anne Joutel** "Pathogenic aspects of Hereditary Small Vessel Disease of the brain" in Cerebral Small Vessel Disease, Cambridge University Press 2014

Patents

- Gene involved in CADASIL, method of diagnosis and therapeutic application. US6995257 (B1)
- Method for the screening of Familial Hemiplegic Migraine. US5714319 (A)
- Immunological treatment of CADASIL. WO2016046053 (A1)

Conferences –Invited Speaker

- Groupe Lillois de Recherche en Pathologie Vasculaire, Lille, 8 May 1997
- Spring Course of the Netherlands Institute for Health Sciences "Advances in Epidemiology and Genetics of Neurologic Diseases", Rotterdam (Netherlands), 31 March 1999
- Groupe Paris Nord de Recherche en Pathologie Vasculaire, Paris, 2000
- Forum of European NeuroScience, Brighton (UK), 28 June 2000
- World Alzheimer Congress 2000, Washington (USA), 2000
- Department of Genetics, University of Cambridge, Cambridge (UK), 23 November 2000
- Fondation IPSEN, XVIIeme Colloque Médecine et Recherche sur la maladie d'Alzheimer, Paris, 2001
- European Neurological Society, ***Anita Harding Lecture***, Paris, 2001
- Société de Biologie "Les nouvelles maladies neurodégénératives", Paris, 2001
- Paris ; Mini colloque IFR6-FR14; Circulation Paris 7, Paris, 2002
- 7th European Stroke Summer School, Graissy en France, 7-13 July 2003
- Gordon Conference "Angiogenesis and Microcirculation", Newport (USA), 10-15 August 2003
- Dept of Microbiology and Molecular Genetics, University of Vermont, Burlington (USA), 16 September 2003
- Third International Congress on Vascular Dementia, Prague (Czechoslovakia), 23-26 October 2003
- 9th Annual meeting of the European Council Cardiovascular Research, Nice, 1-3 October 2004
- 7° colloque de la Société des NeuroSciences, Lille, 17-20 May 2005
- German Society of Neurogenetics, Münster (Germany), 8-10 September 2005
- Cantoblanco Workshop on Biology « Notch signaling in vertebrate development and disease », Madrid (Spain), 21-24 May 2006
- Third CADASIL symposium , Stockholm (Sueden), 30 October-1 November 2006
- The Notch meeting, Athènes (Greece), 23-27 September 2007
- Le Club des Belles Souris, Institut Curie, Orsay, 15 November 2007
- 42° Congrès du collège Français de Pathologie vasculaire, Paris, 12-14 March 2008
- Colloque du Collège de France « Actualités dans le domaine des maladies monogéniques : mécanismes physiopathologiques, approches thérapeutiques », Paris, 15-16 April 2008
- 26° GRRC, Nancy, 2-3 April 2009
- NAVBO meeting « Genetics and Genomics of Vascular Diseases », CapCod (USA), 13-16 September 2009
- 6th International congress on Vascular Dementia, Barcelone (Spain), 19-22 November
- Groupe Paris Nord Circulation, Paris, 2 December 2009
- Ludwig-Maximilians-Universität München, Munich (Germany), 2010
- United leukodystrophy Foundation « CADASIL meeting », Dekalb, Illinois (USA), 2010
- 5th Congress of the International Society for Vascular Behavioural and Cognitive Disorders (VAS-COG), Lille, 11-14 September 2011
- XX European Stroke conference, Hambourg (Germany), 24-27 May 2011
- Totman Guest Lecturer for the Department of Pharmacology's Annual Research Retreat, Burlington (USA), 26-28 October 2011
- Société Cerveau et Maladies cérébrovasculaires, Paris, 20 January 2012
- International Stroke Conference 2012, New Orleans (USA), February 2012
- CADASIL conference, Salt Lake City (USA), 12-14 September 2012

- Special Symposium on Vascular Biology, Ludwig-Maximilians-University, Munich (Germany), 14-15 March 2013
- 54th Annual Meeting of the Japanese Society of Neurology, Tokyo (Japan), 29 May-1 June 2013
- Second International Workshop on CADASIL, Fondation Singer-Polignac, Paris, 19-20 September 2013
- Danish Society for Neuroscience, Sandbjerg (Denmark), 4-6 May 2014
- 9th International Research Symposium on Marfan Syndrome and Related Disorders, Paris, 25-27 September 2014
- Anne McLaren Laboratory for Regenerative Medicine, Cambridge (UK), 18 September 2014
- 14th Eibsee-Meeting "Cellular Mechanisms of Neurodegeneration", Munich (Germany), 5-7 November 2014
- European Stroke conference, Glasgow (UK), 16-19 April 2015
- AVIESAN, Workshop "The neurovascular unit in health and disease", Paris, 26 May 2015
- MRC Centre for Regenerative Medicine Edinburgh (UK), 28 September 2015
- 9th International congress on vascular dementia, Lubiana (Solevnia), 16-18 October 2015
- European Stroke Organization workshop, Garmisch-Patenkirchen (Germany), 19-21 November 2015
- Journées Recherche & Santé – Accident Vasculaire Cérébral, de la clinique à la physiopathologie Paris, 24 November 2015
- 25th European Stroke Conference, Venise (Italy), 13-15 April 2016)
- Federation of European Physiological Societies Congress 2016, Paris, 29 June-1 July 2016
- FASEB meeting Smooth muscle cells, Lisbon (Portugal), 18-22 July 2016
- 19th international Vascular Biology Meeting, Boston (USA), 30 October -3 November 2016
- British Heart Foundation Glasgow Cardiovascular Research Centre, Glasgow (UK), 24 November 2016
- Brain ischemia & Stroke 16 conference, Rome (Italy), 7-9 December 2016
- Workshop Small vessels, dementia and chronic diseases, Glasgow (UK), 26-27 January 2017
- British Neuropathological Society, Symposium on Small Vessel Diseases, London (UK), 1 March 2017
- Printemps de la Cardiologie 2017, Nantes, 6 April 2017
- Novonordisk Foundation Symposium on Small Vessel Diseases, Coppenhague (Denmark), 11-12 May 2017
- The Alzheimer's Association International Conference® 2017 satellite meeting "Vascular Factors in Dementia and Neurodegeneration", Londres (UK) 13-14 July 2017,
- Zilkha Neurogenetic Institute, Keck School of Medicine of the University of Southern California, Los Angeles (USA), 12-13 October 2017