**Supplementary data 1**

1. Abou Al-Shaar, H., Qadi, N., Al-Hamed, M. H., Meyer, B. F., & Bohlega, S. (2016). Phenotypic comparison of individuals with homozygous or heterozygous mutation of NOTCH3 in a large CADASIL family. Journal of the Neurological Sciences, 367, 239–243. https://doi.org/10.1016/j.jns.2016.05.061
2. Abramycheva, N., Stepanova, M., Kalashnikova, L., Zakharova, M., Maximova, M., Tanashyan, M., … Illarioshkin, S. (2015). New mutations in the Notch3 gene in patients with cerebral autosomal dominant arteriopathy with subcortical infarcts and leucoencephalopathy (CADASIL). Journal of the Neurological Sciences, 349(1–2), 196–201. https://doi.org/10.1016/j.jns.2015.01.018
3. Adib-Samii, P., Brice, G., Martin, R. J., & Markus, H. S. (2010). Clinical spectrum of CADASIL and the effect of cardiovascular risk factors on phenotype: Study in 200 consecutively recruited individuals. Stroke, 41(4), 630–634. https://doi.org/10.1161/STROKEAHA.109.568402
4. Alexander, S. K., Brown, J. M., Graham, A., & Nestor, P. J. (2014). CADASIL presenting with a behavioural variant frontotemporal dementia phenotype. Journal of Clinical Neuroscience: Official Journal of the Neurosurgical Society of Australasia, 21(1), 165–167. https://doi.org/10.1016/j.jocn.2013.02.025
5. Andreadou, E., Papadimas, G., & Sfagos, C. (2008). A novel heterozygous mutation in the NOTCH3 gene causing CADASIL. Swiss Medical Weekly, 138(41–42), 614–617. https://doi.org/2008/41/smw-12394
6. Arboleda-Velasquez, J. F., Lopera, F., Lopez, E., Frosch, M. P., Sepulveda-Falla, D., Gutierrez, J. E., … Kosik, K. S. (2002). C455R notch3 mutation in a Colombian CADASIL kindred with early onset of stroke. Neurology, 59(2), 277–279.
7. Arboleda-Velasquez, J. F., Lopera, F., Lopez, E., Frosch, M. P., Sepulveda-Falla, D., Gutierrez, J. E., … Kosik, K. S. (2002). C455R notch3 mutation in a Colombian CADASIL kindred with early onset of stroke. Neurology, 59(2), 277–279. https://doi.org/10.1212/wnl.59.2.277
8. Au, K.-M., Li, H.-L., Sheng, B., Chow, T.-C., Chen, M.-L., Lee, K.-C., & Chan, A. Y.-W. (2007). A novel mutation (C271F) in the Notch3 gene in a Chinese man with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Clinica Chimica Acta; International Journal of Clinical Chemistry, 376(1–2), 229–232. https://doi.org/10.1016/j.cca.2006.07.022
9. Benabu, Y., Beland, M., Ferguson, N., Maranda, B., & Boucher, R.-M. (2013). Genetically proven cerebral autosomal-dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) in a 3-year-old. Pediatric Radiology, 43(9), 1227–1230. https://doi.org/10.1007/s00247-013-2658-5
10. Bentley, P., Wang, T., Malik, O., Nicholas, R., Ban, M., Sawcer, S., & Sharma, P. (2011). CADASIL with cord involvement associated with a novel and atypical NOTCH3 mutation. Journal of Neurology, Neurosurgery, and Psychiatry, 82(8), 855–860. https://doi.org/10.1136/jnnp.2010.223297
11. Bersano, A., Bedini, G., Markus, H. S., Vitali, P., Colli-Tibaldi, E., Taroni, F., … Lombardia GENS-group. (2018). The role of clinical and neuroimaging features in the diagnosis of CADASIL. Journal of Neurology, 265(12), 2934–2943. https://doi.org/10.1007/s00415-018-9072-8
12. Bersano, A., Ranieri, M., Ciammola, A., Cinnante, C., Lanfranconi, S., Dotti, M. T., … Bassi, M. T. (2013). Considerations on a mutation in the NOTCH3 gene sparing a cysteine residue: A rare polymorphism rather than a CADASIL variant. Functional Neurology, 27(4), 247–252.
13. Bianchi, S., Rufa, A., Ragno, M., D’Eramo, C., Pescini, F., Pantoni, L., … Federico, A. (2010). High frequency of exon 10 mutations in the NOTCH3 gene in Italian CADASIL families: Phenotypic peculiarities. Journal of Neurology, 257(6), 1039–1042. https://doi.org/10.1007/s00415-010-5481-z
14. Bianchi, S., Zicari, E., Carluccio, A., Di Donato, I., Pescini, F., Nannucci, S., … Dotti, M. T. (2015). CADASIL in central Italy: A retrospective clinical and genetic study in 229 patients. Journal of Neurology, 262(1), 134–141. https://doi.org/10.1007/s00415-014-7533-2
15. Bianchi S., D. M. (n.d.). First deep intronic mutation in the NOTCH3 gene in a family with late onset CADASIL. - PubMed—NCBI. Retrieved March 7, 2019,
16. Bohlega, S., Al Shubili, A., Edris, A., Alreshaid, A., AlKhairallah, T., AlSous, M. W., … Abu-Amero, K. K. (2007). CADASIL in Arabs: Clinical and genetic findings. BMC Medical Genetics, 8, 67. https://doi.org/10.1186/1471-2350-8-67
17. Brass, S. D., Smith, E. E., Arboleda-Velasquez, J. F., Copen, W. A., & Frosch, M. P. (2009). Case records of the Massachusetts General Hospital. Case 12-2009. A 46-year-old man with migraine, aphasia, and hemiparesis and similarly affected family members. The New England Journal of Medicine, 360(16), 1656–1665. https://doi.org/10.1056/NEJMcpc0810839
18. Buczek, J., Błażejewska-Hyżorek, B., Cudna, A., Lusawa, M., Lewandowska, E., Kurkowska-Jastrzębska, I., & Członkowska, A. (2016). Novel mutation of the NOTCH3 gene in a Polish family with CADASIL. Neurologia I Neurochirurgia Polska, 50(4), 262–264. https://doi.org/10.1016/j.pjnns.2016.04.008
19. Cappelli, A., Ragno, M., Cacchiò, G., Scarcella, M., Staffolani, P., & Pianese, L. (2009). High recurrence of the R1006C NOTCH3 mutation in central Italian patients with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). Neuroscience Letters, 462(2), 176–178. https://doi.org/10.1016/j.neulet.2009.06.087
20. Ceroni, M., Poloni, T. E., Tonietti, S., Fabozzi, D., Uggetti, C., Frediani, F., … Carrera, P. (2000). Migraine with aura and white matter abnormalities: Notch3 mutation. Neurology, 54(9), 1869–1871.
21. Chabriat, H., Vahedi, K., Iba-Zizen, M. T., Joutel, A., Nibbio, A., Nagy, T. G., … Ducrocq, X. (1995). Clinical spectrum of CADASIL: A study of 7 families. Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Lancet (London, England), 346(8980), 934–939.
22. Chan, T. L. H., Sharma, M., & Burneo, J. G. (2018). Focal Epilepsy Secondary to Juxtacortical Lesions in Cerebral Autosomal Dominant Arteriopathy with Subacute Infarcts and Leukoencephalopathy. The Canadian Journal of Neurological Sciences. Le Journal Canadien Des Sciences Neurologiques, 45(4), 462–463. https://doi.org/10.1017/cjn.2018.29
23. Chen, B. S., Cleland, J., King, R. I., & Anderson, N. E. (2019). CADASIL presenting with focal and generalised epilepsy due to a novel NOTCH3 mutation. Seizure, 66, 36–38. https://doi.org/10.1016/j.seizure.2019.01.026
24. Chen, S., Ni, W., Yin, X.-Z., Liu, H.-Q., Lu, C., Zheng, Q.-J., … Wu, Z.-Y. (2017). Clinical features and mutation spectrum in Chinese patients with CADASIL: A multicenter retrospective study. CNS Neuroscience & Therapeutics, 23(9), 707–716. https://doi.org/10.1111/cns.12719
25. Choi, J. C., Lee, K.-H., Song, S.-K., Lee, J. S., Kang, S.-Y., & Kang, J.-H. (2013). Screening for NOTCH3 gene mutations among 151 consecutive Korean patients with acute ischemic stroke. Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 22(5), 608–614. https://doi.org/10.1016/j.jstrokecerebrovasdis.2011.10.013
26. Choi, J. C., Song, S.-K., Lee, J. S., Kang, S.-Y., & Kang, J.-H. (2013). Diversity of stroke presentation in CADASIL: Study from patients harboring the predominant NOTCH3 mutation R544C. Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 22(2), 126–131. https://doi.org/10.1016/j.jstrokecerebrovasdis.2011.07.002
27. Choi, J. C., Song, S.-K., Lee, J. S., Kang, S.-Y., & Kang, J.-H. (2014). Headache among CADASIL patients with R544C mutation: Prevalence, characteristics, and associations. Cephalalgia: An International Journal of Headache, 34(1), 22–28. https://doi.org/10.1177/0333102413497598
28. Chuah, T. L., Tan, K. M., Tan, S. M., Flanagan, S., Hyland, V., Sullivan, A. A., … Lander, C. (2001). CADASIL (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leucoencephalopathy): An Australian perspective. Journal of Clinical Neuroscience: Official Journal of the Neurosurgical Society of Australasia, 8(5), 404–406. https://doi.org/10.1054/jocn.2000.0848
29. Cima, A.-N., Fica, S. V., Albu, A. I., & Lambrescu, I. M. (n.d.). Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy in acromegalic patient with severe headache. 5.
30. Cleves, C., Friedman, N. R., Rothner, A. D., & Hussain, M. S. (2010). Genetically confirmed CADASIL in a pediatric patient. Pediatrics, 126(6), e1603-1607. https://doi.org/10.1542/peds.2010-0714
31. Collongues, N., Derache, N., Blanc, F., Labauge, P., de Seze, J., & Defer, G. (2012). Inflammatory-like presentation of CADASIL: A diagnostic challenge. BMC Neurology, 12, 78. https://doi.org/10.1186/1471-2377-12-78
32. Coto, E., Menéndez, M., Navarro, R., García-Castro, M., & Alvarez, V. (2006). A new de novo Notch3 mutation causing CADASIL. European Journal of Neurology, 13(6), 628–631. https://doi.org/10.1111/j.1468-1331.2006.01337.x
33. Cumurciuc, R., Massin, P., Paques, M., Krisovic, V., Gaudric, A., Bousser, M., & Chabriat, H. (2004). Retinal abnormalities in CADASIL: A retrospective study of 18 patients. Journal of Neurology, Neurosurgery, and Psychiatry, 75(7), 1058–1060. https://doi.org/10.1136/jnnp.2003.024307
34. de la Peña, P., Bornstein, B., del Hoyo, P., Fernández-Moreno, M. A., Martín, M. A., Campos, Y., … Garesse, R. (2001). Mitochondrial dysfunction associated with a mutation in the Notch3 gene in a CADASIL family. Neurology, 57(7), 1235–1238.
35. DE LANGE, R. P. J., BOLT, J., REID, E., DA SILVA, R., SHAW, D., & CLAIR, D. (2000). Screening British CADASIL families for mutations in the NOTCH3 gene. Journal of Medical Genetics, 37(3), 224–225. https://doi.org/10.1136/jmg.37.3.224
36. De Silva, K. R. D., Gamage, R., Dunuwille, J., Gunarathna, D., Sirisena, D., Weerasinghe, A., … Mizuno, T. (2009). Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL): A patient from Sri Lanka. Journal of Clinical Neuroscience: Official Journal of the Neurosurgical Society of Australasia, 16(11), 1492–1493. https://doi.org/10.1016/j.jocn.2009.01.019
37. Delibas, S., Guven, H., & Comoglu, S. S. (2009). A case report about CADASlL: Mutation in the NOTCH 3 receptor. Acta Neurologica Taiwanica, 18(4), 262–266.
38. Desmond, D. W., Moroney, J. T., Lynch, T., Chan, S., Chin, S. S., Shungu, D. C., … Mohr, J. P. (1998). CADASIL in a North American family: Clinical, pathologic, and radiologic findings. Neurology, 51(3), 844–849.
39. Di Donato, I., Bianchi, S., De Stefano, N., Dichgans, M., Dotti, M. T., Duering, M., … Federico, A. (2017). Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) as a model of small vessel disease: Update on clinical, diagnostic, and management aspects. BMC Medicine, 15. https://doi.org/10.1186/s12916-017-0778-8
40. Dichgans, M., Herzog, J., & Gasser, T. (2001). NOTCH3 mutation involving three cysteine residues in a family with typical CADASIL. Neurology, 57(9), 1714–1717.
41. Dichgans, M., Ludwig, H., Müller-Höcker, J., Messerschmidt, A., & Gasser, T. (2000). Small in-frame deletions and missense mutations in CADASIL: 3D models predict misfolding of Notch3 EGF-like repeat domains. European Journal of Human Genetics: EJHG, 8(4), 280–285. https://doi.org/10.1038/sj.ejhg.5200460
42. Dichgans, M., Mayer, M., Uttner, I., Brüning, R., Müller-Höcker, J., Rungger, G., … Gasser, T. (1998). The phenotypic spectrum of CADASIL: Clinical findings in 102 cases. Annals of Neurology, 44(5), 731–739. https://doi.org/10.1002/ana.410440506
43. Domínguez-Sánchez, F. J., Lasa-Aristu, A., & Goñi-Imízcoz, M. (2011). Intelligence impairment, personality features and psychopathology disturbances in a family affected with CADASIL. The Spanish Journal of Psychology, 14(2), 936–943.
44. Dotti, M. T., De Stefano, N., Bianchi, S., Malandrini, A., Battisti, C., Cardaioli, E., & Federico, A. (2004). A novel NOTCH3 frameshift deletion and mitochondrial abnormalities in a patient with CADASIL. Archives of Neurology, 61(6), 942–945. https://doi.org/10.1001/archneur.61.6.942
45. Dotti, M., Federico, A., Mazzei, R., Bianchi, S., Scali, O., Conforti, F., … Quattrone, A. (2005). The spectrum of Notch3 mutations in 28 Italian CADASIL families. Journal of Neurology, Neurosurgery, and Psychiatry, 76(5), 736–738. https://doi.org/10.1136/jnnp.2004.048207
46. Ducray, F., Ritzenthaler, T., Cho, T. H., Bruyas, A., Cotton, F., Cartalat-Carel, S., … Nighoghossian, N. (2010). Acute headache followed by focal neuropsychological impairment in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 19(1), 75–76. https://doi.org/10.1016/j.jstrokecerebrovasdis.2009.03.001
47. Dziewulska, D., Sulejczak, D., & Wężyk, M. (2017). What factors determine phenotype of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)? Considerations in the context of a novel pathogenic R110C mutation in the NOTCH3 gene. Folia Neuropathologica, 55(4), 295–300. https://doi.org/10.5114/fn.2017.72387
48. Ebihara, Y., Mochizuki, H., Ishii, N., Mizuta, I., Shiomi, K., Mizuno, T., & Nakazato, M. (2018). A Japanese Case of CADASIL with a Rare Mutation in Exon 24 of the NOTCH3 Gene. Internal Medicine, 57(20), 3011–3014. https://doi.org/10.2169/internalmedicine.0723-17
49. Ejaz, R., Qin, W., Huang, L., Blaser, S., Tetreault, M., Hartley, T., … Care4Rare Canada Consortium. (2016). Lateral meningocele (Lehman) syndrome: A child with a novel NOTCH3 mutation. American Journal of Medical Genetics. Part A, 170A(4), 1070–1075. https://doi.org/10.1002/ajmg.a.37541
50. Engelter, S. T., Rueegg, S., Kirsch, E. C., Fluri, F., Probst, A., Steck, A. J., & Lyrer, P. A. (2002). CADASIL mimicking primary angiitis of the central nervous system. Archives of Neurology, 59(9), 1480–1483.
51. Fernández, A., Gómez, J., Alonso, B., Iglesias, S., & Coto, E. (2015). A Next-Generation Sequencing of the NOTCH3 and HTRA1 Genes in CADASIL Patients. Journal of Molecular Neuroscience: MN, 56(3), 613–616. https://doi.org/10.1007/s12031-015-0560-3
52. Ferrante, E., Mosca, L., Erminio, C., Penco, S., & Cavallari, U. (2019). Identification of a novel NOTCH3 mutation in an Italian family affected by a mild form of CADASIL. Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology. https://doi.org/10.1007/s10072-019-03774-x
53. Feuerhake, F., Volk, B., Ostertag, C. B., Jungling, F. D., Kassubek, J., Orszagh, M., & Dichgans, M. (2002). Reversible coma with raised intracranial pressure: An unusual clinical manifestation of CADASIL. Acta Neuropathologica, 103(2), 188–192. https://doi.org/10.1007/s004010100439
54. Finnilä, S., Tuisku, S., Herva, R., & Majamaa, K. (2001). A novel mitochondrial DNA mutation and a mutation in the Notch3 gene in a patient with myopathy and CADASIL. Journal of Molecular Medicine (Berlin, Germany), 79(11), 641–647. https://doi.org/10.1007/s001090100268
55. Finsterer, J. (2007). Neuromuscular implications in CADASIL. Cerebrovascular Diseases (Basel, Switzerland), 24(5), 401–404. https://doi.org/10.1159/000108428
56. Furby, A., Vahedi, K., Force, M., Larrouy, S., Ruchoux, M. M., Joutel, A., & Tournier-Lasserve, E. (1998). Differential diagnosis of a vascular leukoencephalopathy within a CADASIL family: Use of skin biopsy electron microscopy study and direct genotypic screening. Journal of Neurology, 245(11), 734–740.
57. Ge, W., Kuang, H., Wei, B., Bo, L., Xu, Z., Xu, X., … Sun, M. (2014). A Novel Cysteine-Sparing NOTCH3 Mutation in a Chinese Family with CADASIL. PLoS ONE, 9(8). https://doi.org/10.1371/journal.pone.0104533
58. Generation and characterization of the human iPSC line IDISi001-A isolated from blood cells of a CADASIL patient carrying a NOTCH3 mutation | Elsevier Enhanced Reader. (n.d.). https://doi.org/10.1016/j.scr.2018.01.023
59. Goate, A. M., & Morris, J. C. (1997). Notch3 mutations and the potential for diagnostic testing for CADASIL. The Lancet, 350(9090), 1490. https://doi.org/10.1016/S0140-6736(97)22047-7
60. Gong, M., Rueschendorf, F., Marx, P., Schulz, H., Kraft, H.-G., Huebner, N., & Koennecke, H.-C. (2010). Clinical and genetic features in a family with CADASIL and high lipoprotein (a) values. Journal of Neurology, 257(8), 1240–1245. https://doi.org/10.1007/s00415-010-5496-5
61. Granild‐Jensen, J., Jensen, U. B., Schwartz, M., & Hansen, U. S. (2009). Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy resulting in stroke in an 11-year-old male. Developmental Medicine & Child Neurology, 51(9), 754–757. https://doi.org/10.1111/j.1469-8749.2008.03241.x
62. Grigg, R., Lea, R., Sullivan, A. A., Curtain, R., MacMillian, J., & Griffiths, L. (2000). Identification of a novel mutation C144F in the Notch3 gene in an Australian CADASIL pedigree. Human Mutation, 16(5), 449–450. https://doi.org/10.1002/1098-1004(200011)16:5<449::AID-HUMU26>3.0.CO;2-I
63. Guerreiro, R. J., Lohmann, E., Kinsella, E., Bras, J. M., Luu, N., Gurulian, N., … Singleton, A. (2012). Exome sequencing reveals an unexpected genetic cause of disease: NOTCH3 mutation in a Turkish family with Alzheimer’s disease. Neurobiology of Aging, 33(5), 1008.e17-1008.e23. https://doi.org/10.1016/j.neurobiolaging.2011.10.009
64. Guerrot, D., François, A., Boffa, J.-J., Boulos, N., Hanoy, M., Legallicier, B., … Godin, M. (2008). Nephroangiosclerosis in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: Is NOTCH3 mutation the common culprit? American Journal of Kidney Diseases: The Official Journal of the National Kidney Foundation, 52(2), 340–345. https://doi.org/10.1053/j.ajkd.2008.04.017
65. Guidetti, D., Casali, B., Mazzei, R. L., Cenacchi, G., De Berti, G., Zuccoli, G., … Brini, M. (2004). An Italian case of CADASIL with mutation CGC-TCG in codon 1006, exon 19 Notch3 gene. Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 24(6), 401–406. https://doi.org/10.1007/s10072-003-0196-x
66. Haan, J., Lesnik Oberstein, S. A. J., & Ferrari, M. D. (2007). Epilepsy in Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy. Cerebrovascular Diseases, 24(2–3), 316–317. https://doi.org/10.1159/000106518
67. Haddad, N., Ikard, C., Hiatt, K., Shanmugam, V., & Schmidley, J. (2015). Recurrent status epilepticus as the primary neurological manifestation of CADASIL: A case report. Epilepsy & Behavior Case Reports, 3, 26–29. https://doi.org/10.1016/j.ebcr.2015.02.004
68. Haritunians, T., Chow, T., De Lange, R. P. J., Nichols, J., Ghavimi, D., Dorrani, N., … Schanen, C. (2005). Functional analysis of a recurrent missense mutation in Notch3 in CADASIL. Journal of Neurology, Neurosurgery, and Psychiatry, 76(9), 1242–1248. https://doi.org/10.1136/jnnp.2004.051854
69. Hartley, J., Westmacott, R., Decker, J., Shroff, M., & Yoon, G. (2010). Childhood-onset CADASIL: Clinical, imaging, and neurocognitive features. Journal of Child Neurology, 25(5), 623–627. https://doi.org/10.1177/0883073810361382
70. He, D., Chen, D., Li, X., Hu, Z., Yu, Z., Wang, W., & luo, X. (2016). The comparisons of phenotype and genotype between CADASIL and CADASIL-like patients and population-specific evaluation of CADASIL scale in China. The Journal of Headache and Pain, 17. https://doi.org/10.1186/s10194-016-0646-5
71. Hou, X., He, C., Jin, Q., Niu, Q., Ren, G., & Cheng, H. (2017). Novel Mutation of the NOTCH3 Gene in a Chinese Pedigree with CADASIL. CNS & Neurological Disorders Drug Targets, 16(1), 30–35. https://doi.org/10.2174/1871527315666161024125952
72. Hsiao, C.-T., Chen, Y.-C., Liu, Y.-T., Soong, B.-W., & Lee, Y.-C. (2015). Acute simultaneous multiple lacunar infarcts as the initial presentation of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Journal of the Chinese Medical Association: JCMA, 78(7), 424–426. https://doi.org/10.1016/j.jcma.2015.01.007
73. Hsieh, I.-C., Kuan, T.-S., Hsieh, P.-C., Chen, S.-M., Yen, W.-J., Chang, W.-C., … Lin, Y.-C. (2014). Detection of early cognitive impairment using AD8 in a young patient with stroke with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy syndrome: A case report. American Journal of Alzheimer’s Disease and Other Dementias, 29(2), 133–137. https://doi.org/10.1177/1533317513511289
74. Hung, L. Y., Ling, T. K., Lau, N. K. C., Cheung, W. L., Chong, Y. K., Sheng, B., … Mak, C. M. (2018). Genetic diagnosis of CADASIL in three Hong Kong Chinese patients: A novel mutation within the intracellular domain of NOTCH3. Journal of Clinical Neuroscience: Official Journal of the Neurosurgical Society of Australasia, 56, 95–100. https://doi.org/10.1016/j.jocn.2018.06.050
75. Ince, B., Benbir, G., Siva, A., Saip, S., Utku, U., Celik, Y., … Uyguner, O. (2014). Clinical and radiological features in CADASIL and NOTCH3-negative patients: A multicenter study from Turkey. European Neurology, 72(3–4), 125–131. https://doi.org/10.1159/000360530
76. Ishida, C., Sakajiri, K., Yoshita, M., Joutel, A., Cave-Riant, F., & Yamada, M. (2006). CADASIL with a Novel Mutation in Exon 7 of NOTCH3 (C388Y). Internal Medicine, 45(16), 981–985. https://doi.org/10.2169/internalmedicine.45.1692
77. Ito, D., Tanahashi, N., Murata, M., Sato, H., Saito, I., Watanabe, K., & Fukuuchi, Y. (2002). Notch3 gene polymorphism and ischaemic cerebrovascular disease. Journal of Neurology, Neurosurgery, and Psychiatry, 72(3), 382–384. https://doi.org/10.1136/jnnp.72.3.382
78. Iwatsuki, K., Murakami, T., Manabe, Y., Narai, H., Warita, H., Hayashi, T., & Abe, K. (2001). Two cases of Japanese CADASIL with corpus callosum lesion. The Tohoku Journal of Experimental Medicine, 195(2), 135–140.
79. Joutel, A., Chabriat, H., Vahedi, K., Domenga, V., Vayssière, C., Ruchoux, M. M., … Tournier-Lasserve, E. (2000). Splice site mutation causing a seven amino acid Notch3 in-frame deletion in CADASIL. Neurology, 54(9), 1874–1875.
80. Joutel, A., Corpechot, C., Ducros, A., Vahedi, K., Chabriat, H., Mouton, P., … Tournier-Lasserve, E. (1997). Notch3 mutations in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL), a mendelian condition causing stroke and vascular dementia. Annals of the New York Academy of Sciences, 826, 213–217.
81. Joutel, A., Dodick, D. D., Parisi, J. E., Cecillon, M., Tournier-Lasserve, E., & Bousser, M. G. (2000). De novo mutation in the Notch3 gene causing CADASIL. Annals of Neurology, 47(3), 388–391.
82. Joutel, A., Favrole, P., Labauge, P., Chabriat, H., Lescoat, C., Andreux, F., … Tournier-Lasserve, E. (2001). Skin biopsy immunostaining with a Notch3 monoclonal antibody for CADASIL diagnosis. Lancet (London, England), 358(9298), 2049–2051. https://doi.org/10.1016/S0140-6736(01)07142-2
83. Joutel, Anne, Andreux, F., Gaulis, S., Domenga, V., Cecillon, M., Battail, N., … Tournier-Lasserve, E. (2000). The ectodomain of the Notch3 receptor accumulates within the cerebrovasculature of CADASIL patients. Journal of Clinical Investigation, 105(5), 597–605.
84. Joutel, Anne, Corpechot, C., Ducros, A., Vahedi, K., Chabriat, H., Mouton, P., … Tournier-Lasserve, E. (1996). Notch3 mutations in CADASIL, a hereditary adult-onset condition causing stroke and dementia. Nature, 383(6602), 707–710. https://doi.org/10.1038/383707a0
85. Joutel, Anne, Monet, M., Domenga, V., Riant, F., & Tournier-Lasserve, E. (2004). 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. American Journal of Human Genetics, 74(2), 338–347.
86. Joutel, Anne, Vahedi, K., Corpechot, C., Troesch, A., Chabriat, H., Vayssière, C., … Tournier-Lasserve, E. (1997). Strong clustering and stereotyped nature of Notch3 mutations in CADASIL patients. The Lancet, 350(9090), 1511–1515. https://doi.org/10.1016/S0140-6736(97)08083-5
87. Kamimura, K., Takahashi, K., Uyama, E., Tokunaga, M., Kotorii, S., Uchino, M., & Tabira, T. (1999). Identification of a Notch3 mutation in a Japanese CADASIL family. Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Alzheimer Disease and Associated Disorders, 13(4), 222–225.
88. Kilarski, L. L., Rutten-Jacobs, L. C. A., Bevan, S., Baker, R., Hassan, A., Hughes, D. A., & Markus, H. S. (2015). Prevalence of CADASIL and Fabry Disease in a Cohort of MRI Defined Younger Onset Lacunar Stroke. PLoS ONE, 10(8). https://doi.org/10.1371/journal.pone.0136352
89. Kim, H.-J., Kim, H. Y., Paek, W. K., Park, A., Young Park, M., Ki, C. S., … Kim, S. H. (2012). Amyotrophic lateral sclerosis and frontotemporal lobar degeneration in association with CADASIL. The Neurologist, 18(2), 92–95. https://doi.org/10.1097/NRL.0b013e318247bb2d
90. Kim, Y., Choi, E. J., Choi, C. G., Kim, G., Choi, J. H., Yoo, H. W., & Kim, J. S. (2006). Characteristics of CADASIL in Korea: A novel cysteine-sparing Notch3 mutation. Neurology, 66(10), 1511–1516. https://doi.org/10.1212/01.wnl.0000216259.99811.50
91. Kim, Y.-E., Yoon, C. W., Seo, S. W., Ki, C.-S., Kim, Y. B., Kim, J.-W., … Na, D. L. (2014). Spectrum of NOTCH3 mutations in Korean patients with clinically suspicious cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Neurobiology of Aging, 35(3), 726.e1-6. https://doi.org/10.1016/j.neurobiolaging.2013.09.004
92. Kim, Youngho, Kim, J. S., Kim, G., No, Y. J., & Yoo, H.-W. (2006). Two novel mutations of the NOTCH3 gene in Korean patients with CADASIL. Mutation Research, 593(1–2), 116–120. https://doi.org/10.1016/j.mrfmmm.2005.06.031
93. Kobayashi, J., Sato, S., Okumura, K., Miyashita, F., Ueda, A., Ando, Y., & Toyoda, K. (2014). Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy without anterior temporal pole involvement: A case report. Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 23(3), e241-242. https://doi.org/10.1016/j.jstrokecerebrovasdis.2013.10.013
94. Kotorii, S., Takahashi, K., Kamimura, K., Nishio, T., Arima, K., Yamada, H., … Tabira, T. (2001). Mutations of the notch3 gene in non-caucasian patients with suspected CADASIL syndrome. Dementia and Geriatric Cognitive Disorders, 12(3), 185–193. https://doi.org/10.1159/000051256
95. Koutroulou, I., Karapanayiotides, T., Grigoriadis, N., & Karacostas, D. (2016). CADASIL presenting with spontaneous intracerebral hemorrhage: Report of a case and description of the first family in Northern Greece. Hippokratia, 20(1), 76–79.
96. La Piana, R., Leppert, I. R., Pike, G. B., Lanthier, S., Brais, B., & Tampieri, D. (2018). 3T MRI study discloses high intrafamilial variability in CADASIL due to a novel NOTCH3 mutation. Journal of Clinical Neuroscience: Official Journal of the Neurosurgical Society of Australasia, 58, 25–29. https://doi.org/10.1016/j.jocn.2018.10.080
97. Lackovic, V., Bajcetic, M., Lackovic, M., Novakovic, I., Labudović Borović, M., Pavlovic, A., … Kostic, V. (2012). Skin and sural nerve biopsies: Ultrastructural findings in the first genetically confirmed cases of CADASIL in Serbia. Ultrastructural Pathology, 36(5), 325–335. https://doi.org/10.3109/01913123.2012.679352
98. Lågas, P. A., & Juvonen, V. (2001). Schizophrenia in a patient with cerebral autosomally dominant arteriopathy with subcortical infarcts and leucoencephalopathy (CADASIL disease). Nordic Journal of Psychiatry, 55(1), 41–42. https://doi.org/10.1080/080394801750093724
99. Lee, J. S., Kang, C., Park, S. Q., Choi, H. A., & Sim, K.-B. (2015). Clinical Significance of Cerebral Microbleeds Locations in CADASIL with R544C NOTCH3 Mutation. PLoS ONE, 10(2). https://doi.org/10.1371/journal.pone.0118163
100. Lee, S. J., Meng, H., Elmadhoun, O., Blaivas, M., & Wang, M. M.-H. (2011). Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy affecting an African American man: Identification of a novel 15-base pair NOTCH3 duplication. Archives of Neurology, 68(12), 1584–1586. https://doi.org/10.1001/archneurol.2011.781
101. Lee, Y.-C., Liu, C.-S., Chang, M.-H., Lin, K.-P., Fuh, J.-L., Lu, Y.-C., … Soong, B.-W. (2009). Population-specific spectrum of NOTCH3 mutations, MRI features and founder effect of CADASIL in Chinese. Journal of Neurology, 256(2), 249–255. https://doi.org/10.1007/s00415-009-0091-3
102. Lee, Y.-C., Yang, A.-H., & Soong, B.-W. (2009). The remarkably variable expressivity of CADASIL: Report of a minimally symptomatic man at an advanced age. Journal of Neurology, 256(6), 1026–1027. https://doi.org/10.1007/s00415-009-5048-z
103. Lee, Y.-C., Yang, A.-H., Liu, H.-C., Wong, W.-J., Lu, Y.-C., Chang, M.-H., & Soong, B.-W. (2006). Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: Two novel mutations in the NOTCH3 gene in Chinese. Journal of the Neurological Sciences, 246(1–2), 111–115. https://doi.org/10.1016/j.jns.2006.02.011
104. Lesnik Oberstein, S. A. J., van Duinen, S. G., van den Boom, R., Maat-Schieman, M. L. C., van Buchem, M. A., van Houwelingen, H. C., … Haan, J. (2003). Evaluation of diagnostic NOTCH3 immunostaining in CADASIL. Acta Neuropathologica, 106(2), 107–111. https://doi.org/10.1007/s00401-003-0701-6
105. Li, S., Chen, Y., Shan, H., Ma, F., Shi, M., & Xue, J. (2017). Novel heterozygous NOTCH3 pathogenic variant found in two Chinese patients with CADASIL. Journal of Clinical Neuroscience: Official Journal of the Neurosurgical Society of Australasia, 46, 85–89. https://doi.org/10.1016/j.jocn.2017.08.029
106. Liao, Y.-C., Hsiao, C.-T., Fuh, J.-L., Chern, C.-M., Lee, W.-J., Guo, Y.-C., … Lee, Y.-C. (2015). Characterization of CADASIL among the Han Chinese in Taiwan: Distinct Genotypic and Phenotypic Profiles. PLoS ONE, 10(8). https://doi.org/10.1371/journal.pone.0136501
107. Liem, M. K., Lesnik Oberstein, S. A. J., Vollebregt, M. J., Middelkoop, H. A. M., van der Grond, J., & Helderman-van den Enden, A. T. J. M. (2008). Homozygosity for a NOTCH3 mutation in a 65-year-old CADASIL patient with mild symptoms: A family report. Journal of Neurology, 255(12), 1978–1980. https://doi.org/10.1007/s00415-009-0036-x
108. Lim, K.-S., Tan, A.-H., Lim, C.-S., Chua, K.-H., Lee, P.-C., Ramli, N., … Ng, C.-C. (2015). R54C Mutation of NOTCH3 Gene in the First Rungus Family with CADASIL. PLoS ONE, 10(8). https://doi.org/10.1371/journal.pone.0135470
109. Liu, X., Zuo, Y., Sun, W., Zhang, W., Lv, H., Huang, Y., … Wang, Z. (2015). The genetic spectrum and the evaluation of CADASIL screening scale in Chinese patients with NOTCH3 mutations. Journal of the Neurological Sciences, 354(1–2), 63–69. https://doi.org/10.1016/j.jns.2015.04.047
110. Low, W. C., Junna, M., Börjesson-Hanson, A., Morris, C. M., Moss, T. H., Stevens, D. L., … Kalaria, R. N. (2007). Hereditary multi-infarct dementia of the Swedish type is a novel disorder different from NOTCH3 causing CADASIL. Brain: A Journal of Neurology, 130(Pt 2), 357–367. https://doi.org/10.1093/brain/awl360
111. Lv, H., Yao, S., Zhang, W., Wang, Z., Huang, Y., Niu, X., … Yuan, Y. (2004). [Clinical features in 4 Chinese families with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)]. Beijing Da Xue Xue Bao. Yi Xue Ban = Journal of Peking University. Health Sciences, 36(5), 496–500.
112. Machowska-Sempruch, K., Bajer-Czajkowska, A., Makarewicz, K., Zaryczańska, K., Koryzma, A., & Nowacki, P. (2019). A Novel NOTCH3 Gene Mutation in a Polish CADASIL Family. Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 28(3), 574–576. https://doi.org/10.1016/j.jstrokecerebrovasdis.2018.10.040
113. Maksemous, N., Smith, R. A., Haupt, L. M., & Griffiths, L. R. (2016). Targeted next generation sequencing identifies novel NOTCH3 gene mutations in CADASIL diagnostics patients. Human Genomics, 10. https://doi.org/10.1186/s40246-016-0093-z
114. Mandellos, D., Limbitaki, G., Papadimitriou, A., & Anastasopoulos, D. (2005). Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) in a Greek family. Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 26(4), 278–281. https://doi.org/10.1007/s10072-005-0472-z
115. Martikainen, M. H., & Roine, S. (2012). Rapid improvement of a complex migrainous episode with sodium valproate in a patient with CADASIL. The Journal of Headache and Pain, 13(1), 95–97. https://doi.org/10.1007/s10194-011-0400-y
116. Matsushima, T., Conedera, S., Tanaka, R., Li, Y., Yoshino, H., Funayama, M., … Hattori, N. (2017). Genotype-phenotype correlations of cysteine replacement in CADASIL. Neurobiology of Aging, 50, 169.e7-169.e14. https://doi.org/10.1016/j.neurobiolaging.2016.10.026
117. Mazzei, R., Conforti, F. L., Lanza, P. L., Sprovieri, T., Lupo, M. R., Gallo, O., … Quattrone, A. (2004). A novel Notch3 gene mutation not involving a cysteine residue in an Italian family with CADASIL. Neurology, 63(3), 561–564.
118. Mehta, S., Mehndiratta, P., & Sila, C. A. (2013). Spontaneous cerebellar hemorrhage associated with a novel Notch3 mutation. Journal of Clinical Neuroscience: Official Journal of the Neurosurgical Society of Australasia, 20(7), 1034–1036. https://doi.org/10.1016/j.jocn.2012.11.003
119. Mellies, J., Calabrese, P., Roth, H., & Gehlen, W. (1999). CADASIL. Fortschritte der Neurologie · Psychiatrie, 67(09), 426–433. https://doi.org/10.1055/s-2007-994992
120. Miranda, M., Dichgans, M., Slachevsky, A., Urbina, F., Mena, I., Venegas, P., & Galvez, M. (2006). CADASIL presenting with a movement disorder: A clinical study of a Chilean kindred. Movement Disorders: Official Journal of the Movement Disorder Society, 21(7), 1008–1012. https://doi.org/10.1002/mds.20879
121. Mizuno, T., Muranishi, M., Torugun, T., Tango, H., Nagakane, Y., Kudeken, T., … Nakagawa, M. (2008). Two Japanese CADASIL Families Exhibiting Notch3 Mutation R75P Not Involving Cysteine Residue. Internal Medicine, 47(23), 2067–2072. https://doi.org/10.2169/internalmedicine.47.1391
122. Moccia, M., Mosca, L., Erro, R., Cervasio, M., Allocca, R., Vitale, C., … Penco, S. (2015). Hypomorphic NOTCH3 mutation in an Italian family with CADASIL features. Neurobiology of Aging, 36(1), 547.e5-11. https://doi.org/10.1016/j.neurobiolaging.2014.08.021
123. Monet-Leprêtre, M., Bardot, B., Lemaire, B., Domenga, V., Godin, O., Dichgans, M., … Joutel, A. (2009). Distinct phenotypic and functional features of CADASIL mutations in the Notch3 ligand binding domain. Brain, 132(6), 1601–1612. https://doi.org/10.1093/brain/awp049
124. Moon, S.-Y., Kim, H.-Y., Seok, J.-I., Kwon, J.-C., Ki, C.-S., Kim, J.-W., … Na, D. L. (2003). A novel mutation (C67Y)in the NOTCH3 gene in a Korean CADASIL patient. Journal of Korean Medical Science, 18(1), 141–144.
125. Moreton, F. C., Razvi, S. S. M., Davidson, R., & Muir, K. W. (2014). Changing clinical patterns and increasing prevalence in CADASIL. Acta Neurologica Scandinavica, 130(3), 197–203. https://doi.org/10.1111/ane.12266
126. Morroni, M., Marzioni, D., Ragno, M., Di Bella, P., Cartechini, E., Pianese, L., … Scarpelli, M. (2013). Role of Electron Microscopy in the Diagnosis of Cadasil Syndrome: A Study of 32 Patients. PLoS ONE, 8(6). https://doi.org/10.1371/journal.pone.0065482
127. Mosca, L., Marazzi, R., Ciccone, A., Santilli, I., Bersano, A., Sansone, V., … Penco, S. (2011). NOTCH3 gene mutations in subjects clinically suspected of CADASIL. Journal of the Neurological Sciences, 307(1–2), 144–148. https://doi.org/10.1016/j.jns.2011.04.019
128. Mosca, L., Rivieri, F., Tanel, R., Bonfante, A., Burlina, A., Manfredini, E., … Penco, S. (2014). Mutational screening of NOTCH3 gene reveals two novel mutations: Complexity of CADASIL diagnosis. Journal of Molecular Neuroscience: MN, 54(4), 723–729. https://doi.org/10.1007/s12031-014-0311-x
129. Muiño, E., Gallego-Fabrega, C., Cullell, N., Carrera, C., Torres, N., Krupinski, J., … Fernández-Cadenas, I. (2017). Systematic Review of Cysteine-Sparing NOTCH3 Missense Mutations in Patients with Clinical Suspicion of CADASIL. International Journal of Molecular Sciences, 18(9). https://doi.org/10.3390/ijms18091964
130. Mukai, M., Mizuta, I., Ueda, A., Nakashima, D., Kushimura, Y., Noto, Y.-I., … Mizuno, T. (2018). A Japanese CADASIL patient with homozygous NOTCH3 p.Arg544Cys mutation confirmed pathologically. Journal of the Neurological Sciences, 394, 38–40. https://doi.org/10.1016/j.jns.2018.08.029
131. Murakami, T., Iwatsuki, K., Hayashi, T., Sato, K., Matsubara, E., Nagano, I., … Abe, K. (2001). Two Japanese CADASIL families with a R141C mutation in the Notch3 gene. Internal Medicine (Tokyo, Japan), 40(11), 1144–1148.
132. Mykkänen, K., Junna, M., Amberla, K., Bronge, L., Kääriäinen, H., Pöyhönen, M., … Viitanen, M. (2009). Different clinical phenotypes in monozygotic CADASIL twins with a novel NOTCH3 mutation. Stroke, 40(6), 2215–2218. https://doi.org/10.1161/STROKEAHA.108.528661
133. Mykkänen, K., Savontaus, M.-L., Juvonen, V., Sistonen, P., Tuisku, S., Tuominen, S., … Pöyhönen, M. (2004). Detection of the founder effect in Finnish CADASIL families. European Journal of Human Genetics, 12(10), 813–819. https://doi.org/10.1038/sj.ejhg.5201221
134. Nakamura, T., Watanabe, H., Hirayama, M., Inukai, A., Kabasawa, H., Matsubara, M., … Sobue, G. (2005). CADASIL with NOTCH3 S180C presenting anticipation of onset age and hallucinations. Journal of the Neurological Sciences, 238(1–2), 87–91. https://doi.org/10.1016/j.jns.2005.07.001
135. Narayan, S. K., Gorman, G., Kalaria, R. N., Ford, G. A., & Chinnery, P. F. (2012). The minimum prevalence of CADASIL in northeast England. Neurology, 78(13), 1025–1027. https://doi.org/10.1212/WNL.0b013e31824d586c
136. Oberstein, S. A., Ferrari, M. D., Bakker, E., van Gestel, J., Kneppers, A. L., Frants, R. R., … Haan, J. (1999). Diagnostic Notch3 sequence analysis in CADASIL: Three new mutations in Dutch patients. Dutch CADASIL Research Group. Neurology, 52(9), 1913–1915.
137. Oki, K., Nagata, E., Ishiko, A., Shimizu, A., Tanaka, K., Takahashi, K., … Suzuki, N. (2007). Novel mutation of the Notch3 gene in a Japanese patient with CADASIL. European Journal of Neurology, 14(4), 464–466. https://doi.org/10.1111/j.1468-1331.2007.01641.x
138. Oliveri, R. L., Muglia, M., De Stefano, N., Mazzei, R., Labate, A., Conforti, F. L., … Quattrone, A. (2001). A novel mutation in the Notch3 gene in an Italian family with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: Genetic and magnetic resonance spectroscopic findings. Archives of Neurology, 58(9), 1418–1422. https://doi.org/10.1001/archneur.58.9.1418
139. Onder, H., Kurtcu, K., Arsava, E. M., & Topcuoglu, M. A. (2017). R141C Mutation of NOTCH3 Gene in Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy. Journal of Neurosciences in Rural Practice, 8(2), 301–303. https://doi.org/10.4103/jnrp.jnrp\_496\_16
140. Opherk, C., Peters, N., Herzog, J., Luedtke, R., & Dichgans, M. (2004). Long-term prognosis and causes of death in CADASIL: A retrospective study in 411 patients. Brain: A Journal of Neurology, 127(Pt 11), 2533–2539. https://doi.org/10.1093/brain/awh282
141. Ozaki, K., Irioka, T., Ishikawa, K., & Mizusawa, H. (2015). CADASIL with a novel NOTCH3 mutation (Cys478Tyr). Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 24(3), e61-62. https://doi.org/10.1016/j.jstrokecerebrovasdis.2014.11.022
142. Palomar, F. J., Suárez, A., Franco, E., Carrillo, F., Gil-Néciga, E., & Mir, P. (2013). Abnormal sensorimotor plasticity in CADASIL correlates with neuropsychological impairment. Journal of Neurology, Neurosurgery, and Psychiatry, 84(3), 329–336. https://doi.org/10.1136/jnnp-2012-303960
143. Pantoni, L., Pescini, F., Nannucci, S., Sarti, C., Bianchi, S., Dotti, M. T., … Inzitari, D. (2010). Comparison of clinical, familial, and MRI features of CADASIL and NOTCH3-negative patients. Neurology, 74(1), 57–63. https://doi.org/10.1212/WNL.0b013e3181c7da7c
144. Paraskevas, G. P., Bougea, A., Synetou, M., Vassilopoulou, S., Anagnostou, E., Voumvourakis, K., … Spengos, K. (2014). CADASIL and autoimmunity: Coexistence in a family with the R169C mutation at exon 4 of the NOTCH3 gene. Cerebrovascular Diseases (Basel, Switzerland), 38(4), 302–307. https://doi.org/10.1159/000369000
145. Paraskevas, G. P., Constantinides, V. C., Yapijakis, C., Kararizou, E., Kapaki, E. N., & Bougea, A. (2018). Recognition of Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) in Two Oligosymptomatic Sisters with Low CADASIL Scale Scores and a Venous Dysplasia: Report of a Novel Greek Family. Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 27(9), e191–e195. https://doi.org/10.1016/j.jstrokecerebrovasdis.2018.04.002
146. Park, S., Park, B., Koh, M. K., & Joo, Y. H. (2014). Case report: Bipolar disorder as the first manifestation of CADASIL. BMC Psychiatry, 14, 175. https://doi.org/10.1186/1471-244X-14-175
147. Passos Gregorio, S., Gattaz, W. F., Tavares, H., Kieling, C., Timm, S., Wang, A. G., … Dias-Neto, E. (2006). Analysis of coding-polymorphisms in NOTCH-related genes reveals NUMBL poly-glutamine repeat to be associated with schizophrenia in Brazilian and Danish subjects. Schizophrenia Research, 88(1–3), 275–282. https://doi.org/10.1016/j.schres.2006.06.036
148. Pavlovic, A. M., Dobricic, V., Semnic, R., Lackovic, V., Novakovic, I., Bajcetic, M., & Sternic, N. (2013). A novel Notch3 Gly89Cys mutation in a Serbian CADASIL family. Acta Neurologica Belgica, 113(3), 299–302. https://doi.org/10.1007/s13760-012-0174-2
149. Peisker, T., Musil, L., Hrebicek, M., Vlaskova, H., Cihelkova, I., & Bartos, A. (2013). Clinical spectrum in CADASIL family with a new mutation. Biomedical Papers, 157(4), 379–382. https://doi.org/10.5507/bp.2013.055
150. Pentti A. Lågas Med.lic., V. J. (2001). Schizophrenia in a patient with cerebral autosomally dominant arteriopathy with subcortical infarcts and leucoencephalopathy (CADASIL disease). Nordic Journal of Psychiatry, 55(1), 41–42. https://doi.org/10.1080/080394801750093724
151. Pescini, F., Bianchi, S., Dotti, M. T., Federico, A., Inzitari, D., & Pantoni, L. (2007). First report of a Romanian CADASIL patient following immigration to Italy. European Journal of Neurology, 14(8), e7-8. https://doi.org/10.1111/j.1468-1331.2007.01743.x
152. Pescini, F., Bianchi, S., Salvadori, E., Poggesi, A., Dotti, M. T., Federico, A., … Pantoni, L. (2008). A pathogenic mutation on exon 21 of the NOTCH3 gene causing CADASIL in an octogenarian paucisymptomatic patient. Journal of the Neurological Sciences, 267(1–2), 170–173. https://doi.org/10.1016/j.jns.2007.10.017
153. Pradotto, L., Azan, G., Doriguzzi, C., Valentini, C., & Mauro, A. (2008). Sporadic vascular dementia as clinical presentation of a new missense mutation within exon 7 of NOTCH3 gene. Journal of the Neurological Sciences, 271(1–2), 207–210. https://doi.org/10.1016/j.jns.2008.04.015
154. Pradotto, L., Orsi, L., Daniele, D., Caroppo, P., Lauro, D., Milesi, A., … Mauro, A. (2012). A new NOTCH3 mutation presenting as primary intracerebral haemorrhage. Journal of the Neurological Sciences, 315(1–2), 143–145. https://doi.org/10.1016/j.jns.2011.12.003
155. Qualtieri, A., Ungaro, C., Bagalà, A., Bianchi, S., Pantoni, L., Moccia, M., & Mazzei, R. (2018). Notch3 protein expression in skin fibroblasts from CADASIL patients. Journal of the Neurological Sciences, 390, 121–128. https://doi.org/10.1016/j.jns.2018.04.027
156. Ragno, M., Cacchiò, G., Fabrizi, G. M., Scarcella, M., Silvaggio, F., Cavallaro, T., … Trojano, L. (2007). Clinical presentation of CADASIL in an Italian patient with a rare Gly528Cys exon 10 NOTCH3 gene mutation. Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 28(4), 181–184. https://doi.org/10.1007/s10072-007-0817-x
157. Ragno, M., Fabrizi, G. M., Cacchiò, G., Scarcella, M., Sirocchi, G., Selvaggio, F., … Trojano, L. (2006). Two novel Italian CADASIL families from Central Italy with mutation CGC-TGC at codon 1006 in the exon 19 Notch3 gene. Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 27(4), 252–256. https://doi.org/10.1007/s10072-006-0679-7
158. Ragno, Michele, Berbellini, A., Cacchiò, G., Manca, A., Di Marzio, F., Pianese, L., … De Michele, G. (2013). Parkinsonism is a late, not rare, feature of CADASIL: A study on Italian patients carrying the R1006C mutation. Stroke, 44(4), 1147–1149. https://doi.org/10.1161/STROKEAHA.111.000458
159. Ragno, Michele, Pianese, L., Cacchiò, G., Manca, A., Scarcella, M., Silvestri, S., … Trojano, L. (2012). Multi-organ investigation in 16 CADASIL families from central Italy sharing the same R1006C mutation. Neuroscience Letters, 506(1), 116–120. https://doi.org/10.1016/j.neulet.2011.10.062
160. Ragno, Michele, Pianese, L., Morroni, M., Cacchiò, G., Manca, A., Di Marzio, F., … Trojano, L. (2013). “CADASIL coma” in an Italian homozygous CADASIL patient: Comparison with clinical and MRI findings in age-matched heterozygous patients with the same G528C NOTCH3 mutation. Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 34(11), 1947–1953. https://doi.org/10.1007/s10072-013-1418-5
161. Ragno, Michele, Pianese, L., Pinelli, M., Silvestri, S., Cacchiò, G., Di Marzio, F., … Castaldo, I. (2011). Shorter telomeres in patients with cerebral autosomal dominant arteriopathy and leukoencephalopathy (CADASIL). Neurogenetics, 12(4), 337–343. https://doi.org/10.1007/s10048-011-0298-1
162. Ragno, Michele, Sanguigni, S., Manca, A., Pianese, L., Paci, C., Berbellini, A., … De Michele, G. (2016). Parkinsonism in a pair of monozygotic CADASIL twins sharing the R1006C mutation: A transcranial sonography study. Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 37(6), 875–881. https://doi.org/10.1007/s10072-016-2497-x
163. Razvi, S., Davidson, R., Bone, I., & Muir, K. (2005). The prevalence of cerebral autosomal dominant arteriopathy with subcortical infarcts and leucoencephalopathy (CADASIL) in the west of Scotland. Journal of Neurology, Neurosurgery, and Psychiatry, 76(5), 739–741. https://doi.org/10.1136/jnnp.2004.051847
164. Rein Gustavsen, W., Reinholt, F. P., & Schlosser, A. (2006). Skin biopsy findings and results of neuropsychological testing in the first confirmed cases of CADASIL in Norway. European Journal of Neurology, 13(4), 359–362. https://doi.org/10.1111/j.1468-1331.2006.01243.x
165. Rinnoci, V., Nannucci, S., Valenti, R., Donnini, I., Bianchi, S., Pescini, F., … Pantoni, L. (2013). Cerebral hemorrhages in CADASIL: Report of four cases and a brief review. Journal of the Neurological Sciences, 330(1–2), 45–51. https://doi.org/10.1016/j.jns.2013.04.002
166. Roine, S., Harju, M., Kivelä, T. T., Pöyhönen, M., Nikoskelainen, E., Tuisku, S., … Summanen, P. A. (2006). Ophthalmologic findings in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: A cross-sectional study. Ophthalmology, 113(8), 1411–1417. https://doi.org/10.1016/j.ophtha.2006.03.030
167. Rufa, A., De Stefano, N., Dotti, M. T., Bianchi, S., Sicurelli, F., Stromillo, M. L., … Federico, A. (2004). Acute unilateral visual loss as the first symptom of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Archives of Neurology, 61(4), 577–580. https://doi.org/10.1001/archneur.61.4.577
168. Rutten, J. W., Haan, J., Terwindt, G. M., van Duinen, S. G., Boon, E. M. J., & Lesnik Oberstein, S. A. J. (2014). Interpretation of NOTCH3 mutations in the diagnosis of CADASIL. Expert Review of Molecular Diagnostics, 14(5), 593–603. https://doi.org/10.1586/14737159.2014.922880
169. Rutten, J. W., Van Eijsden, B. J., Duering, M., Jouvent, E., Opherk, C., Pantoni, L., … Lesnik Oberstein, S. A. J. (2019). The effect of NOTCH3 pathogenic variant position on CADASIL disease severity: NOTCH3 EGFr 1-6 pathogenic variant are associated with a more severe phenotype and lower survival compared with EGFr 7-34 pathogenic variant. Genetics in Medicine: Official Journal of the American College of Medical Genetics, 21(3), 676–682. https://doi.org/10.1038/s41436-018-0088-3
170. Saiki, S., Sakai, K., Saiki, M., Kitagawa, Y., Umemori, T., Murata, K., … Hirose, G. (2006). Varicose veins associated with CADASIL result from a novel mutation in the Notch3 gene. Neurology, 67(2), 337–339. https://doi.org/10.1212/01.wnl.0000224758.52970.19
171. Saito, S., Ozaki, A., Takahashi, M., Ito, H., Matsumoto, S., & Tomimoto, H. (2011). Clustering of multifocal cerebral infarctions in CADASIL: A case report. Journal of Neurology, 258(2), 325–327. https://doi.org/10.1007/s00415-010-5727-9
172. Sakiyama, Y., Matsuura, E., Maki, Y., Yoshimura, A., Ando, M., Nomura, M., … Takashima, H. (2018). Peripheral neuropathy in a case with CADASIL: A case report. BMC Neurology, 18. https://doi.org/10.1186/s12883-018-1131-3
173. Samões, R., Alves, J. E., Taipa, R., Silva, J., Melo Pires, M., & Pereira-Monteiro, J. M. (2016). CADASIL: MRI may be normal in the fourth decade of life - a case report. Cephalalgia: An International Journal of Headache, 36(11), 1082–1085. https://doi.org/10.1177/0333102415618613
174. Sano, Y., Shimizu, F., Kawai, M., Omoto, M., Negoro, K., Kurokawa, T., … Kanda, T. (2011a). P.Arg332Cys Mutation of NOTCH3 Gene in Two Unrelated Japanese Families with CADASIL. Internal Medicine, 50(22), 2833–2838. https://doi.org/10.2169/internalmedicine.50.5418
175. Sano, Y., Shimizu, F., Kawai, M., Omoto, M., Negoro, K., Kurokawa, T., … Kanda, T. (2011b). P.Arg332Cys mutation of NOTCH3 gene in two unrelated Japanese families with CADASIL. Internal Medicine (Tokyo, Japan), 50(22), 2833–2838.
176. Santa, Y., Uyama, E., Chui, D. H., Arima, M., Kotorii, S., Takahashi, K., & Tabira, T. (2003). Genetic, clinical and pathological studies of CADASIL in Japan: A partial contribution of Notch3 mutations and implications of smooth muscle cell degeneration for the pathogenesis. Journal of the Neurological Sciences, 212(1–2), 79–84.
177. Sathe, S., DePeralta, E., Pastores, G., & Kolodny, E. H. (2009). Acute confusional migraine may be a presenting feature of CADASIL. Headache, 49(4), 590–596. https://doi.org/10.1111/j.1526-4610.2009.01363.x
178. Schon, F., Martin, R. J., Prevett, M., Clough, C., Enevoldson, T. P., & Markus, H. S. (2003). “CADASIL coma”: An underdiagnosed acute encephalopathy. Journal of Neurology, Neurosurgery & Psychiatry, 74(2), 249–252. https://doi.org/10.1136/jnnp.74.2.249
179. Schröder, J. M., Züchner, S., Dichgans, M., Nagy, Z., & Molnar, M. J. (2005). Peripheral nerve and skeletal muscle involvement in CADASIL. Acta Neuropathologica, 110(6), 587–599. https://doi.org/10.1007/s00401-005-1082-9
180. Schubert, V., Bender, B., Kinzel, M., Peters, N., & Freilinger, T. (2018). A novel frameshift variant in the CADASIL gene NOTCH3: Pathogenic or not? Journal of Neurology, 265(6), 1338–1342. https://doi.org/10.1007/s00415-018-8844-5
181. Siitonen, M., Mykkänen, K., Pescini, F., Rovio, S., Kääriäinen, H., Baumann, M., … Viitanen, M. (2015). APOE and AGT in the Finnish p.Arg133Cys CADASIL population. Acta Neurologica Scandinavica, 132(6), 430–434. https://doi.org/10.1111/ane.12400
182. Song, J.-K., Noh, Y. O., & Lee, J. S. (2014). Cognitive profile of CADASIL patients with R544C Notch3 mutation. European Neurology, 71(5–6), 217–222. https://doi.org/10.1159/000356199
183. Soong, B.-W., Liao, Y.-C., Tu, P.-H., Tsai, P.-C., Lee, I.-H., Chung, C.-P., & Lee, Y.-C. (2013). A homozygous NOTCH3 mutation p.R544C and a heterozygous TREX1 variant p.C99MfsX3 in a family with hereditary small vessel disease of the brain. Journal of the Chinese Medical Association, 76(6), 319–324. https://doi.org/10.1016/j.jcma.2013.03.002
184. Spinicci G, Conti M.. Unusual clinical presentations in subjects carrying novel NOTCH3 gene mutations. - PubMed—NCBI. Retrieved March 7, 2019
185. Suda, S., Okubo, S., Ueda, M., Sowa, K., Abe, A., Aoki, J., … Kimura, K. (2016). A Japanese CADASIL kindred with a novel two-base NOTCH3 mutation. European Journal of Neurology, 23(5), e32-34. https://doi.org/10.1111/ene.12977
186. Taillia, H., Chabriat, H., Kurtz, A., Verin, M., Levy, C., Vahedi, K., … Bousser, M. G. (1998). Cognitive Alterations in Non-Demented CADASIL Patients. Cerebrovascular Diseases, 8(2), 97–101. https://doi.org/10.1159/000015825
187. Tan, Z.-X., Li, F.-F., Qu, Y.-Y., Liu, J., Liu, G.-R., Zhou, J., … Liu, S.-L. (2012). Identification of a Known Mutation in Notch 3 in Familiar CADASIL in China. PLoS ONE, 7(5). https://doi.org/10.1371/journal.pone.0036590
188. Tang, S., Chen, Y., Chi, N., Chen, C., Cheng, Y., Hsieh, F., … Jeng, J. (2018). Prevalence and clinical characteristics of stroke patients with p.R544C NOTCH3 mutation in Taiwan. Annals of Clinical and Translational Neurology, 6(1), 121–128. https://doi.org/10.1002/acn3.690
189. Tang, S.-C., Lee, M.-J., Jeng, J.-S., & Yip, P.-K. (2005). Arg332Cys mutation of NOTCH3 gene in the first known Taiwanese family with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Journal of the Neurological Sciences, 228(2), 125–128.
190. Testi, S., Malerba, G., Ferrarini, M., Ragno, M., Pradotto, L., Mauro, A., & Fabrizi, G. M. (2012). Mutational and haplotype map of NOTCH3 in a cohort of Italian patients with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). Journal of the Neurological Sciences, 319(1–2), 37–41. https://doi.org/10.1016/j.jns.2012.05.025
191. Tojima, M., Saito, S., Yamamoto, Y., Mizuno, T., Ihara, M., & Fukuda, H. (2016). Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy with a Novel NOTCH3 Cys323Trp Mutation Presenting Border-Zone Infarcts: A Case Report and Literature Review. Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 25(8), e128-130. https://doi.org/10.1016/j.jstrokecerebrovasdis.2016.05.013
192. Tuominen, S., Juvonen, V., Amberla, K., Jolma, T., Rinne, J. O., Tuisku, S., … Kalimo, H. (2001). Phenotype of a homozygous CADASIL patient in comparison to 9 age-matched heterozygous patients with the same R133C Notch3 mutation. Stroke, 32(8), 1767–1774.
193. Tuominen, S., Miao, Q., Kurki, T., Tuisku, S., Pöyhönen, M., Kalimo, H., … Rinne, J. O. (2004). Positron emission tomography examination of cerebral blood flow and glucose metabolism in young CADASIL patients. Stroke, 35(5), 1063–1067. https://doi.org/10.1161/01.STR.0000124124.69842.2d
194. Ueda, A., Ueda, M., Nagatoshi, A., Hirano, T., Ito, T., Arai, N., … Ando, Y. (2015). Genotypic and phenotypic spectrum of CADASIL in Japan: The experience at a referral center in Kumamoto University from 1997 to 2014. Journal of Neurology, 262(8), 1828–1836. https://doi.org/10.1007/s00415-015-7782-8
195. Ungaro, C., Servillo, P., Mazzei, R., Consoli, D., Conforti, F. L., Sprovieri, T., … Quattrone, A. (2009). A pathogenic rare mutation on exon 22 of the NOTCH3 gene disclosed in an Italian patient affected by CADASIL. Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 30(3), 269–271. https://doi.org/10.1007/s10072-009-0040-z
196. Unlü, M., de Lange, R. P., de Silva, R., Kalaria, R., & St Clair, D. (2000). Detection of complement factor B in the cerebrospinal fluid of patients with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy disease using two-dimensional gel electrophoresis and mass spectrometry. Neuroscience Letters, 282(3), 149–152.
197. Utku, U., Celik, Y., Uyguner, O., Yüksel-Apak, M., & Wollnik, B. (2002). CADASIL syndrome in a large Turkish kindred caused by the R90C mutation in the Notch3 receptor. European Journal of Neurology, 9(1), 23–28.
198. Uyama, E., Tokunaga, M., Suenaga, A., Kotorii, S., Kamimura, K., Takahashi, K., … Uchino, M. (2000). Arg133Cys mutation of Notch3 in two unrelated Japanese families with CADASIL. Internal Medicine (Tokyo, Japan), 39(9), 732–737.
199. Uyguner, Z. O., Siva, A., Kayserili, H., Saip, S., Altintaş, A., Apak, M. Y., … Wollnik, B. (2006). The R110C mutation in Notch3 causes variable clinical features in two Turkish families with CADASIL syndrome. Journal of the Neurological Sciences, 246(1–2), 123–130. https://doi.org/10.1016/j.jns.2006.02.021
200. Valenti, R., Bianchi, S., Pescini, F., D’Eramo, C., Inzitari, D., Dotti, M. T., & Pantoni, L. (2011). First report of a pathogenic mutation on exon 24 of the NOTCH3 gene in a CADASIL family. Journal of Neurology, 258(9), 1632–1636. https://doi.org/10.1007/s00415-011-5983-3
201. Van der Aa, N., Vandeweyer, G., & Kooy, R. F. (2010). A boy with mental retardation, obesity and hypertrichosis caused by a microdeletion of 19p13.12. European Journal of Medical Genetics, 53(5), 291–293. https://doi.org/10.1016/j.ejmg.2010.05.006
202. Velizarova, R., Mourand, I., Serafini, A., Crespel, A., & Gelisse, P. (2011). Focal epilepsy as first symptom in CADASIL. Seizure, 20(6), 502–504. https://doi.org/10.1016/j.seizure.2011.02.006
203. Viitanen, M., & Kalimo, H. (2000). CADASIL: Hereditary arteriopathy leading to multiple brain infarcts and dementia. Annals of the New York Academy of Sciences, 903, 273–284.
204. Vikelis, M., Papatriantafyllou, J., & Karageorgiou, C. E. (2007). A novel CADASIL-causing mutation in a stroke patient. Swiss Medical Weekly, 137(21–22), 323–325. https://doi.org/2007/21/smw-11816
205. Vinciguerra, C., Rufa, A., Bianchi, S., Sperduto, A., De Santis, M., Malandrini, A., … Federico, A. (2014). Homozygosity and severity of phenotypic presentation in a CADASIL family. Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 35(1), 91–93. https://doi.org/10.1007/s10072-013-1580-9
206. Vishnevetsky, A., Inca-Martinez, M., Milla-Neyra, K., Barrientos-Iman, D. M., Cornejo-Herrera, I., Cosentino, C., & Cornejo-Olivas, M. (2016). The first report of CADASIL in Peru: Olfactory dysfunction on initial presentation. ENeurologicalSci, 5, 15–19. https://doi.org/10.1016/j.ensci.2016.09.001
207. Wang, J., Li, J., Kong, F., Lv, H., & Guo, Z. (2017). Bipolar II disorder as the initial presentation of CADASIL: An underdiagnosed manifestation. Neuropsychiatric Disease and Treatment, 13, 2175–2179. https://doi.org/10.2147/NDT.S142321
208. Wang, T., Sharma, S., Fox, N., Rossor, M., Brown, M., & Sharma, P. (2000). Description of a simple test for CADASIL disease and determination of mutation frequencies in sporadic ischaemic stroke and dementia patients. Journal of Neurology, Neurosurgery, and Psychiatry, 69(5), 652–654. https://doi.org/10.1136/jnnp.69.5.652
209. Wang, Zhao-xia, Lu, H., Zhang, Y., Bu, D., Niu, X., Zhang, Z., … Yuan, Y. (2004). [NOTCH3 gene mutations in four Chinese families with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy]. Zhonghua Yi Xue Za Zhi, 84(14), 1175–1180.
210. Wang, Zhaoxia, Yuan, Y., Zhang, W., Lv, H., Hong, D., Chen, B., … Wu, S. (2011). NOTCH3 mutations and clinical features in 33 mainland Chinese families with CADASIL. Journal of Neurology, Neurosurgery, and Psychiatry, 82(5), 534–539. https://doi.org/10.1136/jnnp.2010.209247
211. Watanabe, M., Adachi, Y., Jackson, M., Yamamoto-Watanabe, Y., Wakasaya, Y., Shirahama, I., … Shoji, M. (2012). An unusual case of elderly-onset cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) with multiple cerebrovascular risk factors. Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 21(2), 143–145. https://doi.org/10.1016/j.jstrokecerebrovasdis.2010.05.008
212. Weiming, F., Yuliang, W., Youjie, L., Xinsheng, L., Shuyang, X., & Zhaoxia, L. (2013). A novel Notch3 deletion mutation in a Chinese patient with cerebral autosomal dominant arteriopathy with subcortical infarcts and leucoencephalopathy (CADASIL). Journal of Clinical Neuroscience: Official Journal of the Neurosurgical Society of Australasia, 20(2), 322–323. https://doi.org/10.1016/j.jocn.2012.02.026
213. Werbrouck, B. F., & De Bleecker, J. L. (2006). Intracerebral haemorrhage in CADASIL. A case report. Acta Neurologica Belgica, 106(4), 219–221.
214. Wollenweber, F. A., Hanecker, P., Bayer-Karpinska, A., Malik, R., Bäzner, H., Moreton, F., … Duering, M. (2015). Cysteine-sparing CADASIL mutations in NOTCH3 show proaggregatory properties in vitro. Stroke, 46(3), 786–792. https://doi.org/10.1161/STROKEAHA.114.007472
215. Yadav, S., Bentley, P., Srivastava, P., Prasad, K., & Sharma, P. (2013). The first Indian-origin family with genetically proven cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 22(1), 28–31. https://doi.org/10.1016/j.jstrokecerebrovasdis.2011.05.023
216. Yeung, W. T. E., Mizuta, I., Watanabe-Hosomi, A., Yokote, A., Koizumi, T., Mukai, M., … Mizuno, T. (2018). RNF213-related susceptibility of Japanese CADASIL patients to intracranial arterial stenosis. Journal of Human Genetics, 63(5), 687–690. https://doi.org/10.1038/s10038-018-0428-9
217. Yin, X., Wu, D., Wan, J., Yan, S., Lou, M., Zhao, G., & Zhang, B. (2015). Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: Phenotypic and mutational spectrum in patients from mainland China. The International Journal of Neuroscience, 125(8), 585–592. https://doi.org/10.3109/00207454.2014.951929
218. Yin, X.-Z., Ding, M.-P., Zhang, B.-R., Liu, J.-R., Zhang, L., Wang, P.-Z., … Zhao, G.-H. (2009). Report of two Chinese families and a review of Mainland Chinese CADASIL patients. Journal of the Neurological Sciences, 279(1–2), 88–92. https://doi.org/10.1016/j.jns.2008.12.011
219. Yoon, C. W., Kim, Y.-E., Seo, S. W., Ki, C.-S., Choi, S. H., Kim, J.-W., & Na, D. L. (2015). NOTCH3 variants in patients with subcortical vascular cognitive impairment: A comparison with typical CADASIL patients. Neurobiology of Aging, 36(8), 2443.e1-7. https://doi.org/10.1016/j.neurobiolaging.2015.04.009
220. You, J., Liao, S., Zhang, F., Ma, Z., & Li, G. (2017). First Report of Arg587Cys Mutation of Notch3 Gene in Two Chinese Families with CADASIL. Journal of Stroke and Cerebrovascular Diseases: The Official Journal of National Stroke Association, 26(1), e1–e4. https://doi.org/10.1016/j.jstrokecerebrovasdis.2016.09.014
221. Zea-Sevilla, M. A., Bermejo-Velasco, P., Serrano-Heranz, R., & Calero, M. (2015). Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) associated with a novel C82R mutation in the NOTCH3 gene. Journal of Alzheimer’s Disease: JAD, 43(2), 363–367. https://doi.org/10.3233/JAD-141218
222. Zhang, L., Yang, X., Wang, Y., & Pei, L. (2009). [The clinical and genetic studies in a family of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy]. Zhonghua Yi Xue Yi Chuan Xue Za Zhi = Zhonghua Yixue Yichuanxue Zazhi = Chinese Journal of Medical Genetics, 26(2), 187–190. https://doi.org/10.3760/cma.j.issn.1003-9406.2009.02.015
223. Zhang, X., Lee, S. J., Young, M. F., & Wang, M. M. (2015). The small leucine-rich proteoglycan BGN accumulates in CADASIL and binds to NOTCH3. Translational Stroke Research, 6(2), 148–155. https://doi.org/10.1007/s12975-014-0379-1
224. Zhu, Y., Wang, J., Wu, Y., Wang, G., & Hu, B. (2015). Two novel mutations in NOTCH3 gene causes cerebral autosomal dominant arteriopathy with subcritical infarct and leucoencephalopathy in two Chinese families. International Journal of Clinical and Experimental Pathology, 8(2), 1321–1327.