

Article original, JL BERNARD, Editeur KJER France : rédigé : 30/01/2015 ; publié : 01/02/2015

# Liste des publications scientifiques concernant les Atrophies Optiques Dominantes Année 2014

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La liste ci-dessous a été établie à partir des publications parues dans la base de données PubMed sous les critères : AOD et OPA1 durant l'année 2014. Les publications rapportant des recherches éloignées de notre centre d'intérêt ont été écartées de cette liste.

Les **numéros en rouge** gras signalent les publications dans lesquelles un auteur, au moins, appartient à un laboratoire de recherche français [voir [8](#), [22](#), [23](#), [31](#)]

Tout au long de l'année les publications nouvelles sont mises à jour chaque semaine sur le site KJER France suivre le lien vers le NCBI ( marge de droite).

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1. [Mitochondrial Ca\(2+\) uptake correlates with the severity of the symptoms in autosomal dominant optic atrophy.](#)

Fülöp L, Rajki A, Maka E, Molnár MJ, Spät A.

Cell Calcium. 2014 Dec 9. doi:pii: S0143-4160(14)00196-1. 10.1016/j.ceca.2014.11.008. [Epub ahead of print]

2. [Disturbed mitochondrial dynamics and neurodegenerative disorders.](#)

Burté F, Carelli V, Chinnery PF, Yu-Wai-Man P.

Nat Rev Neurol. 2015 Jan;11(1):11-24. doi: 10.1038/nrneurol.2014.228. Epub 2014 Dec 9. Review.

3. [A novel mutation in a case of dominant optic atrophy?](#)

Ramkumar HL, Savino PJ.

Indian J Ophthalmol. 2014 Oct;62(10):1034-6. doi: 10.4103/0301-4738.146043. No abstract available.

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4. [Optical coherence tomography shows early loss of the inferior temporal quadrant retinal nerve fiber layer in autosomal dominant optic atrophy.](#)

Park SW, Hwang JM.

Graefes Arch Clin Exp Ophthalmol. 2014 Nov 19. [Epub ahead of print]

5. [A recurrent deletion mutation in OPA1 causes autosomal dominant optic atrophy in a Chinese family.](#)

Zhang L, Shi W, Song L, Zhang X, Cheng L, Wang Y, Ge X, Li W, Zhang W, Min Q, Jin ZB, Qu J, Gu F.

Sci Rep. 2014 Nov 6;4:6936. doi: 10.1038/srep06936.

6. [Genetic Basis of Mitochondrial Optic Neuropathies.](#)

Maresca A, Caporali L, Strobbe D, Zanna C, Malavolta D, La Morgia C, Valentino ML, Carelli V.

Curr Mol Med. 2014 Oct 10. [Epub ahead of print]

7. [OPA1-dependent cristae modulation is essential for cellular adaptation to metabolic demand.](#)

Patten DA, Wong J, Khacho M, Soubannier V, Mailloux RJ, Pilon-Larose K, MacLaurin JG, Park DS, McBride HM, Trinkle-Mulcahy L, Harper ME, Germain M, Slack RS.

EMBO J. 2014 Nov 18;33(22):2676-91. doi: 10.15252/embj.201488349. Epub 2014 Oct 8.

8. [Improved Locus-Specific Database for OPA1 Mutations Allows Inclusion of Advanced Clinical Data.](#)

Ferré M, Caignard A, Milea D, Leruez S, Cassereau J, Chevrollier A, Amati-Bonneau P, Verny C, Bonneau D, Procaccio V, Reynier P.

Hum Mutat. 2015 Jan;36(1):20-5. doi: 10.1002/humu.22703. Epub 2014 Dec 1.

9. [Mutation screening of mitochondrial DNA as well as OPA1 and OPA3 in a Chinese cohort with suspected hereditary optic atrophy.](#)

Chen J, Xu K, Zhang X, Jiang F, Liu L, Dong B, Ren Y, Li Y.

Invest Ophthalmol Vis Sci. 2014 Sep 9;55(10):6987-95. doi: 10.1167/iovs.14-14953.

10. [Mitochondria: from cell death executioners to regulators of cell differentiation.](#)

Kasahara A, Scorrano L.

Trends Cell Biol. 2014 Dec;24(12):761-770. doi: 10.1016/j.tcb.2014.08.005. Epub 2014 Sep 2. Review.

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11. [Macular sensitivity and fixation patterns in patients with autosomal dominant optic atrophy.](#)

Rönnbäck C, Larsen M.

Dan Med J. 2014 Sep;61(9):A4888.

12. [Clinical and molecular genetic findings in autosomal dominant OPA3-related optic neuropathy.](#)

Sergouniotis PI, Perveen R, Thiselton DL, Giannopoulos K, Sarros M, Davies JR, Biswas S, Ansons AM, Ashworth JL, Lloyd IC, Black GC, Votruba M.

Neurogenetics. 2014 Aug 27. [Epub ahead of print]

13. [\[Not only optic neuropathy: new molecular and clinical aspects of OPA1 gene mutations\].](#)

Oldak M, Scieżyńska A, Szulborski K, Szaflik JP, Szaflik J.

Klin Oczna. 2014;116(1):52-8. Review. Polish.

14. [Medical management of hereditary optic neuropathies.](#)

La Morgia C, Carbonelli M, Barboni P, Sadun AA, Carelli V.

Front Neurol. 2014;5:141. doi: 10.3389/fneur.2014.00141. Review.

15. [Mitochondrial dynamic changes in health and genetic diseases.](#)

Chen L, Winger AJ, Knowlton AA.

Mol Biol Rep. 2014 Nov;41(11):7053-62. doi: 10.1007/s11033-014-3663-y.

16. [Pure and syndromic optic atrophy explained by deep intronic OPA1 mutations and an intralocus modifier.](#)

Bonifert T, Karle KN, Tonagel F, Batra M, Wilhelm C, Theurer Y, Schoenfeld C, Kluba T, Kamenisch Y, Carelli V, Wolf J, Gonzalez MA, Speziani F, Schüle R, Züchner S, Schöls L, Wissinger B, Synofzik M.

Brain. 2014 Aug;137(Pt 8):2164-77. doi: 10.1093/brain/awu165. Epub 2014 Jun 25.

17. [Mutant ubiquitin UBB+1 induces mitochondrial fusion by destabilizing mitochondrial fission-specific proteins and confers resistance to oxidative stress-induced cell death in astrocytic cells.](#)

Yim N, Ryu SW, Han EC, Yoon J, Choi K, Choi C.

PLoS One. 2014;9(6):e99937. doi: 10.1371/journal.pone.0099937.

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18. [197th ENMC international workshop: Neuromuscular disorders of mitochondrial fusion and fission - OPA1 and MFN2 molecular mechanisms and therapeutic strategies: 26-28 April 2013, Naarden, The Netherlands.](#)

Yu-Wai-Man P, Carelli V, Chinnery PF.

Neuromuscul Disord. 2014 Aug;24(8):736-42. doi: 10.1016/j.nmd.2014.05.004. Epub 2014 May 21. No abstract available.

19. [Early macular retinal ganglion cell loss in dominant optic atrophy: genotype-phenotype correlation.](#)

Barboni P, Savini G, Cascavilla ML, Caporali L, Milesi J, Borrelli E, La Morgia C, Valentino ML, Triolo G, Lembo A, Carta A, De Negri A, Sadun F, Rizzo G, Parisi V, Pierro L, Bianchi Marzoli S, Zeviani M, Sadun AA, Bandello F, Carelli V.

Am J Ophthalmol. 2014 Sep;158(3):628-36.e3. doi: 10.1016/j.ajo.2014.05.034. Epub 2014 Jun 5.

20. [First report of OPA1 screening in Greek patients with autosomal dominant optic atrophy and identification of a previously undescribed OPA1 mutation.](#)

Kamakari S, Koutsodontis G, Tsilimbaris M, Fitsios A, Chrousos G.

Mol Vis. 2014;20:691-703.

21. [Biosynthesis and roles of phospholipids in mitochondrial fusion, division and mitophagy.](#)

Zhang Q, Tamura Y, Roy M, Adachi Y, Iijima M, Sesaki H.

Cell Mol Life Sci. 2014 Oct;71(19):3767-78. doi: 10.1007/s00018-014-1648-6. Epub 2014 May 28. Review.

22. [Mitochondrial dysfunction affecting visual pathways.](#)

Leruez S, Amati-Bonneau P, Verny C, Reynier P, Procaccio V, Bonneau D, Milea D.

Rev Neurol (Paris). 2014 May;170(5):344-54. doi: 10.1016/j.neurol.2014.03.009. Epub 2014 May 3.

23. [Perspectives of drug-based neuroprotection targeting mitochondria.](#)

Procaccio V, Bris C, Chao de la Barca JM, Oca F, Chevrollier A, Amati-Bonneau P, Bonneau D, Reynier P.

Rev Neurol (Paris). 2014 May;170(5):390-400. doi: 10.1016/j.neurol.2014.03.005. Epub 2014 May 1.

24. [Dietary supplementation with fish oil alters the expression levels of proteins governing mitochondrial dynamics and prevents high-fat diet-induced endothelial dysfunction.](#)

Sun R, Wang X, Liu Y, Xia M.

Br J Nutr. 2014 Jul;112(2):145-53. doi: 10.1017/S0007114514000701. Epub 2014 Apr 28.

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25. [Two novel compound heterozygous mutations in OPA3 in two siblings with OPA3-related 3-methylglutaconic aciduria.](#)

Lam C, Gallo LK, Dineen R, Ciccone C, Dorward H, Hoganson GE, Wolfe L, Gahl WA, Huizing M. Mol Genet Metab Rep. 2014 Jan 1;1:114-123.

26. [Proteolytic cleavage of Opa1 stimulates mitochondrial inner membrane fusion and couples fusion to oxidative phosphorylation.](#)

Mishra P, Carelli V, Manfredi G, Chan DC.

Cell Metab. 2014 Apr 1;19(4):630-41. doi: 10.1016/j.cmet.2014.03.011. Erratum in: Cell Metab. 2014 May 6;19(5):891.

27. [\[Normal skin biopsy as a tool for extra-cutaneous disorders\].](#)

Kluger N, Freitag S, Roguedas AM, Misery L.

Ann Dermatol Venereol. 2014 Mar;141(3):192-200. doi: 10.1016/j.annder.2014.01.005. Epub 2014 Feb 17. Review. French.

28. [Impaired OMA1-dependent cleavage of OPA1 and reduced DRP1 fission activity combine to prevent mitophagy in cells that are dependent on oxidative phosphorylation.](#)

MacVicar TD, Lane JD.

J Cell Sci. 2014 May 15;127(Pt 10):2313-25. doi: 10.1242/jcs.144337. Epub 2014 Mar 14.

29. [The i-AAA protease YME1L and OMA1 cleave OPA1 to balance mitochondrial fusion and fission.](#)

Anand R, Wai T, Baker MJ, Kladt N, Schauss AC, Rugarli E, Langer T.

J Cell Biol. 2014 Mar 17;204(6):919-29. doi: 10.1083/jcb.201308006. Epub 2014 Mar 10.

30. [Retinal vessel diameters decrease with macular ganglion cell layer thickness in autosomal dominant optic atrophy and in healthy subjects.](#)

Rönnbäck C, Grønskov K, Larsen M.

Acta Ophthalmol. 2014 Nov;92(7):670-4. doi: 10.1111/aos.12378. Epub 2014 Mar 11.

**31.** [Author reply: To PMID 24120325.](#)

Rönnbäck C, Milea D, Larsen M.

Ophthalmology. 2014 Jun;121(6):e30-1. doi: 10.1016/j.ophtha.2013.12.040. Epub 2014 Feb 26. No abstract available.

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32. [Re: Rönnbäck et al.: Imaging of the macula indicates early completion of structural deficit in autosomal-dominant optic atrophy \(Ophthalmology 2013;120:2672-7\).](#)

Abegg M, Zinkernagel M, Wolf S.

Ophthalmology. 2014 Jun;121(6):e29-30. doi: 10.1016/j.ophtha.2013.11.044. Epub 2014 Feb 26. No abstract available.

33. [ROMO1 is an essential redox-dependent regulator of mitochondrial dynamics.](#)

Norton M, Ng AC, Baird S, Dumoulin A, Shutt T, Mah N, Andrade-Navarro MA, McBride HM, Sreaton RA.

Sci Signal. 2014 Jan 28;7(310):ra10. doi: 10.1126/scisignal.2004374.

34. [O ROM\(e\)O1, ROM\(e\)O1, wherefore art thou ROM\(e\)O1?](#)

Semenzato M, Scorrano L.

Sci Signal. 2014 Jan 28;7(310):pe2. doi: 10.1126/scisignal.2005024. Review.

35. [A new non-canonical pathway of Gα\(q\) protein regulating mitochondrial dynamics and bioenergetics.](#)

Benincá C, Planagumà J, de Freitas Shuck A, Acín-Perez R, Muñoz JP, de Almeida MM, Brown JH, Murphy AN, Zorzano A, Enríquez JA, Aragay AM.

Cell Signal. 2014 May;26(5):1135-46. doi: 10.1016/j.cellsig.2014.01.009. Epub 2014 Jan 18.

36. [Autosomal dominant hereditary optic neuropathy \(ADOA\): a review of the genetics and clinical manifestations of ADOA and ADOA+.](#)

Skidd PM, Lessell S, Cestari DM.

Semin Ophthalmol. 2013 Sep-Nov;28(5-6):422-6. doi: 10.3109/08820538.2013.825296. Review.

37. [Mitochondrial morphology-emerging role in bioenergetics.](#)

Galloway CA, Lee H, Yoon Y.

Free Radic Biol Med. 2012 Dec 15;53(12):2218-28. doi: 10.1016/j.freeradbiomed.2012.09.035. Epub 2012 Sep 29. Review.

