Liste des publications scientifiques concernant les Atrophies Optiques Dominantes

Année 2013

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 2013. Les publications rapportant des recherches éloignées de notre centre d'intérêt ont été écartées de cette liste.

Dans cet ensemble, nous relevons les articles ou les thèmes suivants :

- Le premier essai clinique avec idébénone dans les AOD [voir notre résumé]
- Le rôle de l'OCT dans le diagnostic des AOD [voir 1, 5, 9, 24]

Les numéros en rouge gras signalent les publications dans lesquelles un auteur, au moins, appartient à un laboratoire de recherche français [voir 4, 6, 7, 14, 17, 18, 37]

Les publications sont présentées de la plus récente à la plus ancienne

1. High-resolution en face images of microcystic macular edema in patients with autosomal dominant optic atrophy.

Gocho K, Kikuchi S, Kabuto T, Kameya S, Shinoda K, Mizota A, Yamaki K, Takahashi H.

Biomed Res Int. 2013;2013:676803. doi: 10.1155/2013/676803. Epub 2013 Nov 28.

Free Article

2. Mitochondrial cristae shape determines respiratory chain supercomplexes assembly and respiratory efficiency.

Cogliati S, Frezza C, Soriano ME, Varanita T, Quintana-Cabrera R, Corrado M, Cipolat S, Costa V, Casarin A, Gomes LC, Perales-Clemente E, Salviati L, Fernandez-Silva P, Enriquez JA, Scorrano L.

Cell. 2013 Sep 26;155(1):160-71. doi: 10.1016/j.cell.2013.08.032. Epub 2013 Sep 19.

3. Membranes in motion: mitochondrial dynamics and their role in apoptosis.

Ugarte-Uribe B, García-Sáez AJ.

Biol Chem. 2013 Nov 2. doi:pii: /j/bchm.just-accepted/hsz-2013-0234/hsz-2013-0234.xml. 10.1515/hsz-2013-0234. [Epub ahead of print]

4. A novel heterozygous OPA3 mutation located in the mitochondrial target sequence results in altered steady-state levels and fragmented mitochondrial network.

Grau T, Burbulla LF, Engl G, Delettre C, Delprat B, Oexle K, Leo-Kottler B, Roscioli T, Krüger R, Rapaport D, Wissinger B, Schimpf-Linzenbold S.

J Med Genet. 2013 Dec;50 (12):848-58. doi: 10.1136/jmedgenet-2013-101774. Epub 2013 Oct 17.

5. Macular spectral domain optical coherence tomography findings in Tanzanian endemic optic neuropathy.

Kisimbi J, Shalchi Z, Mahroo OA, Mhina C, Sanyiwa AJ, Mabey D, Mohamed M, Plant GT.

Brain. 2013 Nov;136(Pt 11):3418-26. doi: 10.1093/brain/awt221. Epub 2013 Sep 9.

6. [Hereditary optic neuropathies: From clinical signs to diagnosis.]

Meunier I, Lenaers G, Hamel C, Defoort-Dhellemmes S.

J Fr Ophtalmol. 2013 Oct 22. doi:pii: S0181-5512(13)00248-9. 10.1016/j.jfo.2013.05.007. [Epub ahead of print] French.

7. Imaging of the Macula Indicates Early Completion of Structural Deficit in Autosomal-Dominant Optic Atrophy.

Rönnbäck C, Milea D, Larsen M.

Ophthalmology. 2013 Oct 10. doi:pii: S0161-6420(13)00724-0. 10.1016/j.ophtha.2013.08.008. [Epub ahead of print]

8. Dominant optic atrophy, OPA1, and mitochondrial quality control: understanding mitochondrial network dynamics.

Alavi MV, Fuhrmann N.

Mol Neurodegener. 2013 Sep 25; 8(1):32. [Epub ahead of print]

9. [Applications of Optical Coherence Tomography (OCT) in Neuro-ophthalmology.]

Kernstock C, Friebe K, Tonagel F.

Klin Monbl Augenheilkd. 2013 Sep 24. [Epub ahead of print] German.

10. First Cases of Dominant Optic Atrophy in Saudi Arabia: Report of Two Novel OPA1 Mutations.
Galvez-Ruiz A, Neuhaus C, Bergmann C, Bolz H.
J Neuroophthalmol. 2013 Sep 18. [Epub ahead of print]

11. 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.

12. Copper deficiency alters cell bioenergetics and induces mitochondrial fusion through up-regulation of MFN2 and OPA1 in erythropoietic cells.
Bustos RI, Jensen EL, Ruiz LM, Rivera S, Ruiz S, Simon F, Riedel C, Ferrick D, Elorza AA.
Biochem Biophys Res Commun. 2013 Aug 2;437(3):426-32. doi: 10.1016/j.bbrc.2013.06.095. Epub 2013 Jul 4.

13. Visual and psychological morbidity among patients with autosomal dominant optic atrophy.
Bailie M, Votruba M, Griffiths PG, Chinnery PF, Yu-Wai-Man P.
Acta Ophthalmol. 2013 Aug;91(5):e413-4. doi: 10.1111/aos.12077. Epub 2013 Mar 4. No abstract available.

14. Why mitochondria must fuse to maintain their genome integrity.
Vidoni S, Zanna C, Rugolo M, Sarzi E, Lenaers G.
Antioxid Redox Signal. 2013 Aug 1;19(4):379-88. doi: 10.1089/ars.2012.4800. Epub 2013 Mar 28.

15. Dominant optic atrophy: novel OPA1 mutations and revised prevalence estimates.
Yu-Wai-Man P, Chinnery PF.
Ophthalmology. 2013 Aug;120(8):1712-1712.e1. doi: 10.1016/j.ophtha.2013.04.022. No abstract available.
Article KJER France

16. Reply: Sensorineural hearing loss in OPA1-linked disorders.
Yu-Wai-Man P, Chinnery PF.
Brain. 2013 Jul;136(Pt 7):e237. doi: 10.1093/brain/aws341. Epub 2013 Feb 4. No abstract available.

17. Sensorineural hearing loss in OPA1-linked disorders.

Leruez S, Milea D, Defoort-Dhellemmes S, Colin E, Crochet M, Procaccio V, Ferré M, Lamblin J, Drouin V, Vincent-Delorme C, Lenaers G, Hamel C, Blanchet C, Juul G, Larsen M, Verny C, Reynier P, Amati-Bonneau P, Bonneau D.

Brain. 2013 Jul;136(Pt 7):e236. doi: 10.1093/brain/aws340. Epub 2013 Feb 4. No abstract available.

Article KJER France

18. TMEM126A is a mitochondrial located mRNA (MLR) protein of the mitochondrial inner membrane.

Hanein S, Garcia M, Fares-Taie L, Serre V, De Keyzer Y, Delaveau T, Perrault I, Delphin N, Gerber S, Schmitt A, Masse JM, Munnich A, Kaplan J, Devaux F, Rozet JM.

Biochim Biophys Acta. 2013 Jun;1830(6):3719-33. doi: 10.1016/j.bbagen.2013.02.025. Epub 2013 Mar 13.

19. Nuclear factors: roles related to mitochondrial deafness.

Luo LF, Hou CC, Yang WX.

Gene. 2013 May 15;520(2):79-89. doi: 10.1016/j.gene.2013.03.041. Epub 2013 Mar 17. Review.

20. Novel OPA1 missense mutation in a family with optic atrophy and severe widespread neurological disorder.

Liskova P, Ulmanova O, Tesina P, Melsova H, Diblik P, Hansikova H, Tesarova M, Votruba M.

Acta Ophthalmol. 2013 May;91(3):e225-31. doi: 10.1111/aos.12038. Epub 2013 Feb 7.

21. New insights into the function and regulation of mitochondrial fission.

Otera H, Ishihara N, Mihara K.

Biochim Biophys Acta. 2013 May;1833(5):1256-68. doi: 10.1016/j.bbamcr.2013.02.002. Epub 2013 Feb 20. Review.

22. OPA1 loss of function affects in vitro neuronal maturation.

Bertholet AM, Millet AM, Guillermin O, Daloyau M, Davezac N, Miquel MC, Belenguer P.

Brain. 2013 May;136(Pt 5):1518-33. doi: 10.1093/brain/awt060. Epub 2013 Mar 29.

23. Genetic and phenotypic variability of optic neuropathies.

Neuhann T, Rautenstrauss B.

Expert Rev Neurother. 2013 Apr;13(4):357-67. doi: 10.1586/ern.13.19. Review.

24. Correlation between visual acuity and OCT-measured retinal nerve fiber layer thickness in a family with ADOA and an OPA1 mutation.
Russo A, Delcassi L, Marchina E, Semeraro F.
Ophthalmic Genet. 2013 Mar-Jun;34(1-2):69-74. doi: 10.3109/13816810.2012.702259. Epub 2012 Jul 11.

25. [Clinical and molecular genetic analysis of hereditary optic neuropathies].
Avetisov SÉ, Sheremet NL, Vorob'eva OK, Eliseeva ÉG, Chukhrova AL, Loginova AN, Khanakova NA, Poliakov AV.
Vestn Oftalmol. 2013 Mar-Apr;129(2):8-13. Russian.

26. N-terminal cleavage of the mitochondrial fusion GTPase OPA1 occurs via a caspase-independent mechanism in cerebellar granule neurons exposed to oxidative or nitrosative stress.
Gray JJ, Zommer AE, Bouchard RJ, Duval N, Blackstone C, Linseman DA.
Brain Res. 2013 Feb 4;1494:28-43. doi: 10.1016/j.brainres.2012.12.001. Epub 2012 Dec 7.

27. Idebenone treatment in patients with OPA1-mutant dominant optic atrophy.
Barboni P, Valentino ML, La Morgia C, Carbonelli M, Savini G, De Negri A, Simonelli F, Sadun F, Caporali L, Maresca A, Liguori R, Baruzzi A, Zeviani M, Carelli V.
Brain. 2013 Feb;136(Pt 2):e231. doi: 10.1093/brain/aws280. Epub 2013 Feb 6. No abstract available.
Article KJER France

28. Loss of OPA1 disturbs cellular calcium homeostasis and sensitizes for excitotoxicity.
Kushnareva YE, Gerencser AA, Bossy B, Ju WK, White AD, Waggoner J, Ellisman MH, Perkins G, Bossy-Wetzel E.
Cell Death Differ. 2013 Feb;20(2):353-65. doi: 10.1038/cdd.2012.128. Epub 2012 Nov 9.

29. Mitochondrial dysfunction in optic neuropathies: animal models and therapeutic options.
Carelli V, La Morgia C, Sadun AA.
Curr Opin Neurol. 2013 Feb;26(1):52-8. doi: 10.1097/WCO.0b013e32835c5f0b. Review.

30. Mitochondrial dynamics in neurodegeneration.
Itoh K, Nakamura K, Iijima M, Sesaki H.
Trends Cell Biol. 2013 Feb;23(2):64-71. doi: 10.1016/j.tcb.2012.10.006. Epub 2012 Nov 16. Review.

31. SDOCT thickness measurements of various retinal layers in patients with autosomal dominant optic atrophy due to OPA1 mutations.
Schild AM, Ristau T, Fricke J, Neugebauer A, Kirchhof B, Sadda SR, Liakopoulos S.
Biomed Res Int. 2013;2013:121398. doi: 10.1155/2013/121398. Epub 2013 Aug 19.

32. Mutation survey of the optic atrophy 1 gene in 193 Chinese families with suspected hereditary optic neuropathy.
Chen Y, Jia X, Wang P, Xiao X, Li S, Guo X, Zhang Q.
Mol Vis. 2013;19:292-302. Epub 2013 Feb 6.

33. Mitochondrial fusion proteins and human diseases.
Ranieri M, Brajkovic S, Riboldi G, Ronchi D, Rizzo F, Bresolin N, Corti S, Comi GP.
Neurol Res Int. 2013;2013:293893. doi: 10.1155/2013/293893. Epub 2013 May 27.

34. Mitochondrial fusion: a mechanism of cisplatin-induced resistance in neuroblastoma cells?
Santin G, Piccolini VM, Barni S, Veneroni P, Giansanti V, Dal Bo V, Bernocchi G, Bottone MG.
Neurotoxicology. 2013 Jan;34:51-60. doi: 10.1016/j.neuro.2012.10.011. Epub 2012 Oct 26.

35. Opa1 is required for proper mitochondrial metabolism in early development.
Rahn JJ, Stackley KD, Chan SS.
PLoS One. 2013;8(3):e59218. doi: 10.1371/journal.pone.0059218. Epub 2013 Mar 14.

36. Non-image-forming light driven functions are preserved in a mouse model of autosomal dominant optic atrophy.
Perganta G, Barnard AR, Katti C, Vachtsevanos A, Douglas RH, MacLaren RE, Votruba M, Sekaran S.
PLoS One. 2013;8(2):e56350. doi: 10.1371/journal.pone.0056350. Epub 2013 Feb 11.

37. The human OPA1delTTAG mutation induces premature age-related systemic neurodegeneration in mouse.
Sarzi E, Angebault C, Seveno M, Gueguen N, Chaix B, Bielicki G, Boddaert N, Mausset-Bonnefont AL, Cazevieille C, Rigau V, Renou JP, Wang J, Delettre C, Brabet P, Puel JL, Hamel CP, Reynier P, Lenaers G.
Brain. 2012 Dec;135(Pt 12):3599-613. doi: 10.1093/brain/aws303.