

BIBLIOGRAPHIE

- AGUIRRE G. : CSNB in the dog (Mol Vis. 1998 Oct 30; 4:23)
- AGUIRRE-HERNANDEZ J : The Finnish lapphund retinal atrophy locus maps to the centromeric region of CFA9. (BMC Vet Res 2007 Jul 10;3:14)
- AHONEN SJ : A CNGB1 Frameshift Mutation in Papillon and Phalène Dogs with Progressive Retinal Atrophy. PLOS One. 2013 Aug 28;8(8): e72122.
- AHONEN SJ : A Novel Missense Mutation in *ADAMTS10* in Norwegian Elkhound Primary Glaucoma. PLoS One. 2014 Nov Vol 9 Issue 11: e111941.
- AHRAM D.F : Identification of genetic loci associated with primary angle-closure glaucoma in the basset hound. (Molecular Vision 2014; 20:497-510).
- AHRAM D.F : Variants in Nebulin (*NEB*) Are Linked to the Development of Familial Primary Angle Closure Glaucoma in Basset Hounds (PLoS One | DOI:10.1371/journal.pone.0126660 May 4, 2015 :01-19)
- ANDRE Catherine : Les tests génétiques dans l'espèce canine (PMCAC 2000 vol.5)
- AUBIN G. : Le génome et la carte génétique (Point Vét. 1996 vol.28)
- BARNETT KC. : Comparative aspects of canine hereditary eye disease. Adv Vet Sci Comp Med 1976; 20: 39-67.
- BARNETT K : Hereditary Cataracte in Dog (J. Small An. Pract. 1978; 19: 109-120)
- BARNETT K : The Diagnosis & Differential Diagnosis of Cataract in Dog (J. Small An. Pract. 1985; 26: 305-316)
- BARNETT K : Autosomal dominant progressive retinal atrophy in Abyssinian Cats (J. Hered. 1985 :76(3) :168-170)
- BUSSE C : Ophthalmic and cone derived electrodiagnostic findings in outbred Miniature Long-haired Dachshunds homozygous for a RPGRIP1 mutation. (Vet Ophthalmol. 2011 May;14(3):146-52)
- CHAUDIEU G. : Affections oculaires héréditaires (Edit. Point Vet. Rueil Malmaison France 2004)
- CHAUDIEU G. : L'anomalie de l'œil du colley et l'œil du chien à robe merle : similarités et différences (PMCAC 2008 ; 43, 109-116)
- CIDECYAN V. : In vivo dynamics of retinal injury and repair in the rhodopsin mutant dog model of human retinitis pigmentosa (PNAS 2005, 102, 5233-5238)
- CLEMENT PJM : Recent advances in understanding the spectrum of canine generalised progressive retinal atrophy (J. Small An Pract 37, 1996)
- CLERC B. : Ophthalmologie vétérinaire (Edit. Point vet. Rueil Malmaison France 1996)
- CURTIS R BARNETT K : Progressive Retinal Atrophy in Miniature Long-Hair Dachshund dog (Br. Vet. J. 1993; 149: 71-85)
- CURTIS R: Late-onset cataract in the Boston terrier. Veterinary Record 1984; 115: 577-578.
- DENIS B. : Les bases de la génétique médicale (Le Point Vét . Rueil Malmaison France 1996 : 28)

- DEKOMIEN G. : (Gener. PRA of Sloughi dogs is due to an 8-bp insertion in exon 21 of the PDE6B gene (Cytogen. Cell. Genet. 90, 2000)
- DEKOMIEN G: Progressive retinal atrophy in Schapendoes dogs: mutation of the newly identified CCDC66 gene. (Neurogenetics. 2010 11: 163-174.)
- DIXON C.J. : Achromatopsia in three sibling Labrador Retrievers in the UK. (Vet Ophthalmol. 2016 Jan;19(1):68-72).
- DOWNS LM : A frameshift mutation in golden retriever dogs with progressive retinal atrophy endorses SLC4A3 as a candidate gene for human retinal degenerations. (PLoS One. 2011;6(6):e21452)
- DOWNS LM : Late-onset progressive retinal atrophy in the Gordon and Irish Setter breeds is associated with a frameshift mutation in C2orf71. Anim Genet. 2012 Jun 12.
- DOWNS LM : Genetic screening for PRA-associated mutations in multiple dog breeds shows that PRA is heterogeneous within and between breeds. Vet Ophthalmol. 2014 Mar;17(2):126-130.
- EKESTEN B : Abnormal Appearance of the Area Centralis in Labrador Retrievers With an ABCA4 Loss-of-function Mutation. Transl Vis Sci Technol. 2022 Feb 1;11(2):36.
- FARIAS FH: An ADAMTS17 splice donor site mutation in dogs with primary lens luxation.(Invest Ophthalmol Vis Sci 2010, 51 :4716-4721)
- FORMAN O P : Parallel mapping and simultaneous sequencing reveals deletions in BCAN and FAM83H associated with discrete inherited disorders in a domestic dog breed. PLoS Genet. 2012 Jan;8(1):e1002462.
- FORMAN OP: Two Independent Mutations in ADAMTS17 Are Associated with Primary Open Angle Glaucoma in the Basset Hound and Basset Fauve de Bretagne Breeds of Dog. PLoS One. 2015 Oct Vol 16.
- FORMAN OP: A Novel Missense Mutation in ADAMTS17 in PBGV Primary Glaucoma. PLoS One. 2015 Oct Vol 16.
- GALIBERT F. et ANDRE C. : Le chien, un modèle pour la génétique des mammifères Médecine/Science n° 8-9, vol. 20, aout-septembre 2004
- GELATT K N : Veterinary Ophthalmology (Third edition 1999)
- GOLDSTEIN O: An ADAM9 mutation in canine cone-rod dystrophy 3 establishes homology with human cone-rod dystrophy 9. (Mol Vis ; 2010 16:1549-1569)
- GOLDSTEIN O: COL9A2 and COL9A3 mutations in canine autosomal recessive ocular skeletal dysplasia.(Mamm Genome. 2010 Aug;21(7-8):398-408)
- GOLDSTEIN O : A non-stop S-antigen gene mutation is associated with late onset hereditary retinal degeneration in dogs. Mol. Vis. 2013; 19:1871-1884.
- GOLDSTEIN O: IQCB1 and PDE6B Mutations Cause Similar Early Onset Retinal Degenerations in Two Closely Related Terrier Dog Breeds. IOVS 2013; 54:7005-7019.
- GORNIK K R : Canine multifocal retinopathy caused by a BEST1 mutation in a Boerboel.. Vet Ophthalmol. 2013 Sep 3.

- GORNIK K R : Canine multifocal retinopathy caused by a BEST1 mutation in a Boerboel.. Vet Ophthalmol (2014) 17,5,368-372)
- GOULD D : ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. Vet Ophthalmol. 2011 Nov;14(6):378-384.
- GRAHN B H : Diagnostic Ophthalmology (Can. Vet. J. 2006 sept vol. 46: 929-930)
- GRAHN B H : Multifocal retinopathy of Great Pyrennes dogs (Can. Vet. J. 2006 sept 47: 491-2)
- GUYON R : Analysis of six candidate genes as potential modifiers of disease expression in canine XLPR1, a model for human X-linked retinitis pigmentosa 3. (Mol.Vis. 2007 Jul 11;13:1094-105.)
- GUZIEWICZ KE : Bestrophine Gene Mutations Cause Canine Multifocal Retinopathy (Inv Ophthalmol & Vis Sci 2007 ;48 :1959-1967)
- GUZIEWICZ KE: Modeling the Structural Consequences of BEST1 Missense Mutations. Adv in Exp Med Biol 2012 Volume 723, 611-618.
- HOFFMANN I: Canine multifocal retinopathy in the Australian Shepherd: a case report. Vet Ophthalmol. 2012 Sep;15 Suppl 2:134-8.
- HUG P. :
- KARLSTAM L : A slowly progressive retinopathy in the Shetland Sheepdog (Congrès de Bruges ESVO 2006).
- KIJAS JM : Canine models of ocular disease (J. of Heredity 2003, 94, 27-30)
- KROPATSCH R: Generalized progressive retinal atrophy in the Irish Glen of Imaal Terrier is associated with a deletion in the ADAM9 gene.(Mol Cell Probes. 2010 Dec;24(6):357-63.)
- KUCHTEY J: Mapping of the disease locus and identification of ADAMTS10 as a candidate gene in a canine model of primary open angle glaucoma. PLoS Genet. 2011 Feb;7(2) : e1001306.
- KUKEKOVA A: Canine RD3 mutation establishes rod cone dysplasia type 2 (rcd2) as ortholog of human and murine rd3. (Mamm Genome; 20(2): 109–123. 2009)
- KUZNETSOVA T: Exclusion of RPGRIP1 ins44 from primary causal association with early-onset cone-rod dystrophy in dogs. Invest Ophthalmol Vis Sci. 2012 Aug 15;53(9):5486-501.
- LEGALL J.Y. : Génétique moléculaire médicale (Cours du CNED 1998)
- LIN C.T : Canine inherited retinal degeneration (J.Small An.Pract.43, 2002)
- LIPPMANN T : Haplotype-defined linkage region for gPRA in Schapendoes dogs (*Molecular Vision* 2007; 13:174-80)
- NARFSTRÖM K : Retinal dystrophy or CSNB in the Briard dog (Vet. Opht. 2, 1999)
- NARFSTROM K : Assessment of hereditary retinal degeneration in the English springer spaniel dog and disease relationship to an RPGRIP1 mutation. 2012;2012:685901
- MENOTTI-RAYMOND M: Mutation in CEP290 discovered for Cat model of human retinal degeneration (J. Hered. 2007 May-Jun;98(3):211-20)
- MENOTTI-RAYMOND M: Widespread retinal degenerative disease mutation (rdAc) discovered among a large number of popular cat breeds.(Vet J. 2010 Oct;186(1):32-38).

MELLERSH CS : Identification of mutations in HSF4 in dogs o three breeds with hereditary cataracts. (Vet. Opht. 2006 sept-oct; 9(5):369-78)

MELLERSH CS : Canine RPGRIP1 mutation establishes cone-rod dystrophy in miniature longhaired dachshund as a homologue of human Leber congenital amaurosis. (Genomics 2006 sept 88(3):293-301)

MELLERSH CS : Mutation in HSF4 Associated with Early but Not Late-Onset Hereditary Cataract in the Boston Terrier (Journal of Heredity 2007:98(5):531–533)

MELLERSH CS : Mutation in HSF4 is associated with hereditary cataract in the Australian Shepherd. Vet Ophthalmol. 2009 Nov-Dec;12(6):372-378.

METALLINOS D. : Canine molecular genetic testing (Vet Clin North Am Sm An Pract ,march 2001)

MITZUKAMI K: Collie eye anomaly in Hokkaido dogs: case study. Vet Ophthalmol. 2012 Mar;15(2):128-32.

MIYADERA K.: Phenotypic variation and genotype-phenotype discordance in canine cone-rod dystrophy with an RPGRIP1 mutation. (Molecular Vision; 15:2287-2305. 2009).

MOSTOSKEY U.V.: Canine molecular genetic diseases (Compend Cont Educ Pract Vet 2000 vol.22)

OLIVER J.A.C : Two Independent Mutations in *ADAMTS17* Are Associated with Primary Open Angle Glaucoma in the Basset Hound and Basset Fauve de Bretagne Breeds of Dog (PloS One October 16, 2015 P 01-14).

OLIVER JAC : Evaluation of *ADAMTS17* in Chinese Shar-Pei with primary open angle glaucoma, primary lens luxation, or both. Am J Vet Res 2018 Jan ;79(1) :98-106.

OLIVER J.A.C. : A variant in *OLFML3* is associated with pectinate ligament abnormality and primary closed-angle glaucoma in Border Collies from the United Kingdom Vet Ophthalmol 2020 Jan;23(1):25-36.

PAGET Sandrine : Diversité génétique dans l'espèce canine (Thèse DEA 2001-2002)

PALANOVA A : Analysis of a deletion in the nephronophthisis 4 gene in different dog breeds. **PLoS One. 2013 Aug +Vet Ophthalmol 2014 Jan.**

PARKER HG: Breed Relationships Facilitate Fine-mapping Studies: a 7.8kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. (Genome Res. 2007 Nov. 17(11): 1562-71.

PETERSEN-JONES SM : cGMP phosphodiesterase-alpha mutation causes PRA in the Cardigan Welsh Corgi dog (Invest. Opht. Vis. Sci. 40, 1999)

PONT RT : A Carbohydrate Sulfotransferase-6 (*CHST6*) gene mutation is associated with Macular Corneal Dystrophy in Labrador Retrievers. Vet Ophthalmol. 2016 Nov ;19(6)488-492

PRIAT Catherine : Cartographie du génome canin (Thèse de doctorat en biologie 1999)

PUGH C.A : Arginine to Glutamine Variant in Olfactomedin Like 3 (*OLFML3*) Is a Candidate for Severe Goniodysgenesis and Glaucoma in the Border Collie Dog Breed. G3 2019 Mar :943-954

SIDJANIN D.J. : Canine *CNGB3* mutations establish cone degeneration as orthologous to the human achromatopsia locus *ACHM3* (Hum. Mol. Gen. 2002)

SOLIGNAC M.: Génétique et évolution (1995)

STADES FC, DJAJADININGRAT-LAANEN SC : Familial non-rcd1 generalised retinal degeneration in Irish Setter (J Small An Pract March 2003)

SUBER ML : Irish setter dogs affected with rod/cone dysplasia contain a nonsense mutation in the rod cGMP phosphodiesterase beta-subunit gene. (Proc Natl Acad Sci U S A. 1993 May 1;90(9):3968-72).

SVENSSON M : Progressive retinal atrophy in the Polski Owczarek Nizinny dog: a clinical and genetic study (*Veterinary Ophthalmology* (2015) 1–11)

TAGU Denis : Principes des techniques de biologie moléculaire (1999)

TANAKA N. : Canine CNGA3 Gene Mutations Provide Novel Insights into Human Achromatopsia-Associated Channelopathies and Treatment. (PLoS One.2015 Sep 25;10(9)

TETAS PONT R : A Carbohydrate Sulfotransferase-6 (*CHST6*) gene mutation is associated with Macular Corneal Dystrophy in Labrador Retrievers (*Veterinary Ophthalmology* (2015) 1–5)

TURNEY C: Pathological and electrophysiological features of a canine cone-rod dystrophy in the miniature longhaired dachshund. (Invest Opht Vis Sci 2007 Sep;48(9):4240-9)

VESKE A. : Retinal dystrophy of S.briard/Briard-beagle dogs (Genomics 57, 1999)

VILBOUX T : Progressive Retinal Atrophy in the Border Collie: A new XLPRA (*BMC Veterinary Research* 2008, 4:10)

WALDE I. : Atlas d'ophtalmologie (Paris Vigot 1990)

WHITING R E H: Multifocal retinopathy in Dachshunds with CLN2 neuronal ceroid lipofuscinosis (Experimental Eye Research 134 (2015) 123–132).

VIK A.C : (Progressive retinal atrophy in Shetland sheepdog is associated with a mutation in the *CNGA1* gene (2015 Stichting International Foundation for Animal Genetics, **46**, 515–521)

WIJK AC : A deletion in nephronophthisis 4 (NPHP4) is associated with recessive cone-rod dystrophy in standard wire-haired dachshund (Gen Res 18:1415-1421, 2008)

WIJK AC : A study of candidate genes for day blindness in the standard wire haired dachshund. BMC Vet Res. 2008 Jul 1;4:23.

WINKLER PA : A large animal model for CNGB1 autosomal recessive retinitis pigmentosa. PLoS One. 2013 Aug 19;8(8):e72229. Papillon et Phalène.

WINKLER PA : A Partial Gene Deletion of *SLC45A2* Causes Oculocutaneous Albinism in Doberman Pinscher Dogs (PLoS One March 2014 | Volume 9 | Issue 3 | e92127)

YEY CY : Genomic deletion of CNGB3 is identical by descent in multiple canine breeds and causes achromatopsia. (BMC Genet. 2013 Apr 20;14:27).

ZANGERL B : Identical mutation in a novel retinal gene cause progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. (Genomics 2006)

ZANGERL B : Assessment of canine BEST1 variations identifies new mutations and establishes an independent bestrophinopathy model (cmr3). Mol. Vis. 2010 Dec. 16 ;16 :2791-804.

ZHANG Q. : Different RPGR exon ORF15 mutations in canids (Hum. Mol. Gen.2002)

ZHANG Q. : Characterization of canine photoreceptor phosducine cDNA (Gene 1998)