Christian Hamel

Born on October 4th, 1955
French

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ACADEMIC TITLES

1999 Competence in medical genetics
1996 Habilitation à Diriger les Recherches (HDR), University Montpellier 1
1994 PhD in Neurosciences, University Montpellier 1
1987 MD, Silver Medal, University Paris XI, physician specialist in Ophthalmology

MEDICAL TRAINING AND DUTIES

2004- Director of the Centre of Reference for Rare Diseases "Diagnosis and Care of Hereditary Sensory Defects", University Hospital, Montpellier
1992- Consultation (1 day/week) for hereditary retinal dystrophy, University Hospital, Montpellier
1981-1987 Internship in medicine and ophthalmology, University Hospital, Caen
1975-1981 Medical studies, Medical School, Rouen

RESEARCH TRAINING AND DUTIES

2003- Director of INSERM Unit 583 (Pathophysiology and therapy of sensory motor defects)
1998- Head of team "Genetics and therapy of retinal blindness"
2000 Nominated "Directeur de Recherches"
1993 Nominated "Chargé de Recherches"
1992-2002 Senior scientist in INSERM Unit 254 (Neurobiology of Audition), director: Rémy Pujol
1987-1991 Post-doctoral fellowship at the National Institute of Health (USA) in the Laboratory of ocular immunopathology (Dr R Nussenblatt), and of Retinal Molecular Biology (Dr G Chader).

TEACHING-EDUCATION

- Professor of the school of Medicine of Montpellier since September 2008
- Montpellier's school of Orthoptia (6 h/year) and university diploma for low vision (2h/year)
- Master programme "Neurobiology and endocrinology" (2h/year)
- Training of 8 guest students, 10 students in DEA, 6 students in PhD, 2 students in MD
- 18 invited lectures in laboratories, 21 in symposia, 39 to public
- Editorial board member of "Ophthalmic Genetics"

EXPERTISES

2003-2007 Vice-president of INSERM committee for the Neurosciences (CSS 08)
1998-2001 Member of the scientific council of Montpellier's medical school

- Referee for 13 thesis and 5 HDR
- Member of ARVO (American association for research in vision and ophthalmology), ISGED (International society for genetic eye disease)
- Member of the scientific council of Fondation de France (Berthe Fouassier committee), Fédération des Aveugles de France, RETINA FRANCE
- Reviewer for: Clinical Genetics, European Journal of Human Genetics, Human Molecular Genetics, Investigative Ophthalmology and Visual Science, Journal of Medical Genetics, Journal of Neuroscience Research, Ophthalmic Genetics

MAIN ACHIEVEMENTS

Discovery and description of the RPE65 protein (M Redmond's lab in USA) and, back to France, finding that mutations cause a particular form of Leber congenital amaurosis. Establishment of a Briard dog colony in collaboration with F Rolling's group (Nantes) to test gene therapy for RPE65 patients in Europe. Discovery that a mitochondrial dynamin-related protein, OPA1, is responsible for the majority of cases of Kjer disease, one of the leading cause of inherited optic neuropathy.

RESEARCH INTERESTS

Finding genes causing inherited diseases of the retina and optic nerve. More specifically

- characterize the retinal diseases due to impairment of the visual cycle genes, and how can we modulate the visual cycle to modify the visual function and the natural history of certain diseases
- why are retinal ganglion cells vulnerable to certain mitochondrial defects and how can we act on mitochondrial functions

Use this knowledge to help patients by providing more accurate informations, better genetic counselling, and by planning strategies of treatment.

SELECTED PUBLICATIONS

1 Hamel CP, Detrick B, Hooks JJ (1990). Evaluation of Ia expression in rat ocular tissue following inoculation with Interferon-gamma. ***Exp Eye Res*** 50, 173-182.

2 Hamel CP, Tsilou E, Pfeffer BA, Hooks JJ, Detrick B, Redmond TM (1993). Molecular cloning and expression of RPE65, a novel retinal pigment epithelium-specific microsomal protein that is post-transcriptionally regulated in vitro. ***J Biol Chem*** 268, 15751-15757.

3 Hamel CP, Jenkins NA, Gilbert DJ, Copeland NG, Redmond TM (1994). The gene for the retinal pigment epithelium-specific protein RPE65 is localized to human 1p31 and mouse 3. ***Genomics*** 20, 509-512.

4 Marlhens F, Bareil C, Griffoin J-M, Zrenner E, Amalric P, Eliaou C, Liu S-Y, Harris E, Redmond TM, Arnaud B, Claustres M, Hamel CP (1997). Mutations in *RPE65* cause Leber's congenital amaurosis. ***Nature Genetics*** 17, 139-141.

5 Marlhens F, Griffoin J-M, Bareil C, Arnaud B, Claustres M, Hamel CP (1998). Autosomal recessive retinal dystrophy associated with two novel mutations in the *RPE65* gene. ***Eur J Hum Genet*** 6, 527-531.

6 Hamel CP, Griffoin J-M, Bazalgette C, Lasquellec L, Duval P-A, Bareil C, Beaufrère L, Bonnet M, Eliaou C, Marlhens F, Schmitt-Bernard CF, Tuffery S, Claustres M, Arnaud B (2000). Génétique moléculaire des rétinopathies pigmentaires : identification de mutations de gènes CHM, RDS, RHO, RPE65, USH2A et XLRS1. ***J Fr Ophtalmol***, 23, 985-995.

7 Delettre C, Lenaers G, Griffoin J-M, Gigarel N, Lorenzo C, Belenguer P, Pelloquin L, Grosgeorges J, Turc-Carel C, Perret E, Astarie-Dequeker C, Lasquellec L, Arnaud B, Ducommun B, Kaplan J, Hamel CP (2000). Nuclear gene *OPA1* encoding a mitochondrial dynamin-related protein is mutated in dominant optic atrophy. ***Nature Genetics*** 26, 207-210.

8 Hamel CP, Griffoin J-M, Laesquellec L, Bazalgette C, Arnaud (2001). Retinal dystrophies caused by mutations in *RPE65*: assessment of visual functions. ***Br J Ophthalmol*** 85, 424-427.

9 Delettre C, Griffoin J-M, Lenaers G, Belenguer P, Hamel CP (2001). Splicing variants and spectrum of mutations in the OPA1 gene. ***Hum Genet*** 109, 584-591.

10 Delprat B, Boulanger A, Wang J, Beaudoin V, Guitton MJ, Ventéo S, Dechesne CJ, Pujol R, Lavigne-Rebillard M, Puel J-L, Hamel CP (2002). Downregulation of otospiralin, a novel inner ear protein, causes hair cell degeneration and deafness. ***J Neurosci*** 22, 1718-1725.

11 Delettre C, Belenguer P, Lenaers G, Hamel CP (2003). Gene structure and chromosomal localization of mouse Opa1: its exclusion from the Bst locus. ***BMC Genet***. 4, 8 Epub.

12 Lavigne-Rebillard M, Delprat B, Surget M-O, Griffoin J-M, Weil D, Arbones M, Vincent R, Hamel CP (2003). Gene structure, chromosomal localization, and mutation screening of the human gene for the inner ear protein otospiralin. ***Neurogenetics*** 4, 137-140.

13 Delprat B, Ruel J, Guitton MJ, Hamard G, Lenoir M, Pujol R, Puel J-L, Brabet P, Hamel CP (2005). Deafness and cochlear fibrocyte alterations in mice deficient for the inner ear protein otospiralin. ***Mol Cell Biol*** 25, 847-843.

14 Kamei S, Chen-Kuo-Chang M, Cazevieille C, Lenaers G, Olichon A, Belenguer P, Roussignol G, Renard N, Eybalin M, Micelin A, Delettre C, Brabet P, Hamel CP (2005). Expression of the Opa1 mitochondrial protein in retinal ganglion cells; its down regulation causes aggregation of the mitochondrial network. ***Invest Ophthalmol Vis Sci***, 46, 4288-4294.

15 Sénéchal A, Humbert G, Surget M-O, Bazalgette C, Bazalgette C, Arnaud B, Arndt C, Laurent E, Brabet P, Hamel CP (2006). Screening genes of the retinoid metabolism: novel *LRAT* mutation in Leber congenital amaurosis. ***Am J Ophthalmol*** 142, 702-704.

16 Hamel C (2006). Retinitis pigmentosa. ***Orphanet J Rare Dis*** Oct 11, 1:40. http://www.OJRD.com/content/1/1/40

17 Ben Salah S, Kamei S, Sénéchal A, Lopez S, Bazalgette C, Bazalgette C, Malrieu-Eliaou C, Zanlonghi X, Hamel CP (2008). Novel *KCNV2* mutations in cone dystrophy with supernormal rod electroretinogram. Am J Ophthalmol, 145, 1099-1106.