CURRICULUM VITAE

Professor Marcela VOTRUBA BM BCh MA PhD FRCOphth

Tel: 00 44 29 2087 0117 **Email:** Votrubam@cf.ac.uk **Nationality:** British

Current position
2014–present: Head of School of Optometry and Vision Sciences, Cardiff University, UK
2011–present: Professor in Ophthalmology, Cardiff University, UK
2003–present: Honorary Consultant Ophthalmologist, University Hospital Wales, Cardiff
Previous positions
2003 - Consultant in Medical Retina, Moorfields Eye Hospital
2002 - Visiting Researcher, National Eye Institute, National Institutes of Health, USA.
Education
2000 PhD, University College, London
2000 Ophthalmology Specialist Register
1992 FRCOphth, Royal College of Ophthalmologists, London
1987 BM BCh (Oxon). Green College, Oxford University
1984 MA Hons 2:1, The Queen's College, Oxford University
Undergraduate Distinctions
1981 Open Scholarship, The Queen's College, Oxford University
1984 First Class Hons. Dissertation, Oxford University
1987 Final Year Clinical Prize: Green College, Green College, Oxford
Postgraduate Prizes
1996 First Prize, The Royal Society of Medicine Ophthalmology Section
Research Fellowships
1995 - 1997 The Guide Dogs for the Blind Association Research Fellowship
1997 - 1999 The Wellcome Trust Vision Research Training Fellowship
1999 - 2001 The Wellcome Trust Advanced Research Fellowship
2002 - 2003 TC Frost Charitable Trust Research Fellowship
2003 - 2007 MRC Clinician Scientist Fellowship
Distinguished Fellowships
2017 Elected Fellow of the Learned Society of Wales
Other Distinctions
2018 - Queen's Anniversary Prize for Higher and further Education, The Royal Anniversary Trust: School of Optometry & Vision Sciences
2017 - Cardiff University School of the Year Award to the School of Optometry & Vision Sciences

Current and recent Research Grants & Fellowships
Total external research funding >**£3.6 million** (>**£3 million as Principal Investigator**)

2013-2015 NIHR Rare Disease Translational Research Collaboration (£200,000). P Yu-Wai-Man, **M Votruba**, AT Moore. "Mitochondrial optic neuropathies".
2013-2016 Fight for Sight Research Studentship (£100,000). **M Votruba,** M Rozanowska. "Light therapy for optic neuropathy".
2013-2014 National Eye Research Centre Project Grant (£60,000). **M Votruba**, M Rozanowska, J Morgan. "Idebenone in mitochondrial optic neuropathy."
2016 EVERf Fellowship **M Votruba**. Research Fellow Dr Deepti Mahajan (£12,000)
2016-2018 Rare Disease Translational Research Collaboration (£200,000). P Yu-Wai-Man, **M Votruba**, "Treatment trial of Epi-743 in mitochondrial optic neuropathies".
2016-2019 ISSF/ Welsh Government/ HEFCW. (£40,000). M Rozanowska, A Brancale, C Heard, **M Votruba**. "Novel drug therapy for mitochondrial optic neuropathy and other mitochondrial diseases."
Welcome Trust ISSF Equipment Award: Pre-clinical OCT: Equipment for development and testing of novel sight-saving therapeutics. (£96,823). **M Votruba**, Co-Applicants

from Cardiff School of Pharmacy and Pharmaceutical Sciences and School of Optometry & Vision Sciences
2016-2019 National Eye Research Centre Project Grant (£60,000). "A novel treatment for blindness caused by autosomal dominant optic atrophy". £60,000. **Votruba M,** Brancale A, Rozanowska M and Heard C
2019-2021 Academy of Medical Sciences Newton International Fellowship. Near-infrared light therapy as a therapeutic regimen for inherited optic neuropathy. £99,000. Host PI Professor **M Votruba**. Fellow Dr Dinesh Kumar Kandaswamy

Other academic activities
National
- 2018- present Invited Member of the James Lind Alliance Priority Setting Partnership Panel: Rare Mitochondrial Diseases
- 2016 - 2018 National Ophthalmology Specialty Lead for Wales & Member of the UK Specialty Lead Panel for Ophthalmology Trials
- 2014 – 2017 Member of the VISION 2020 UK Eye Research Committee
- Lead Principal Investigator and co-coordinator of the MRC Mouse Eyes & Vision Consortium
- 2012 – 2017 Member Steering Committee Eye Genetics Group, UK
- 2012 – 2017 Member Grant Advisory Panel, Fight for Sight, UK
- 2015 – 2020 Member Grant Advisor Panel National Eye Research Centre, UK
- 2017– 2020 Member Grant Advisory Board RP Fighting Blindness, UK
- 2012 – 2013 Royal College of Ophthalmologists Representative on James Lind Alliance Priority Setting Partnership: Sight Loss and Vision Steering Committee

International
- 2008 & 2011 Jury member Belgian Ophthalmology Research Prize
- 2004 & 2009 Jury member College of Ophthalmologists Research Prize
- 2011 – 2016 Programme Secretary European Association for Vision and Eye Research
- 2009 – 2010 Vice President, European Association for Vision and Eye Research
- 2004 – 2009 Section Chair for Molecular Biology/ Genetics/ Epidemiology on the Board of European Association for Vision and Eye Research (EVER)
- Editorial Board of Acta Ophthalmologica,
- Guest Editor for Drug Discovery Today: Disease Models (with Ian Jackson, Edinburgh)
- Since 1998 regularly invited to act as Session Chair at meetings, including EVER (1998/99/2001/05/06/08/09/10) and the XXIXth International Congress of Ophthalmology, Sydney (2002)

Publications
86 refereed journal articles; h-citation index 30 (Google Scholar, i10-index 47, citations 12,825), and h-index 27 (WOS, citations 6543)
Number of papers where the PI is the first or the last (corresponding) author: 47
Number of published book chapters: 3

5 most significant publications
- DJ Klionsky, ….**M Votruba**, ….et al., Guidelines for the use and interpretation of assays for monitoring autophagy. *Autophagy* 2012; **8:** 445-544. **Citations 6830**
- Y Gong, RB Slee, N Fukai, ….**M Votruba** , B Zabel, T Garcia, R Baron, BR Olsen, ML Warman. LDL receptor-related protein 5 (LRP5) affects bone accrual and eye development. *Cell* 2001; **107**: 513-523. **Citation 2121**
- C Alexander, **M Votruba**, U E A Pesch, D L Thiselton, S Mayer, A T Moore, M Rodriguez, U Kellner, B Leo-Kottler, G Auburger, S S Bhattacharya, B Wissinger. A

gene encoding a dynamin-related GTPase is mutated in autosomal dominant optic atrophy. *Nature Genetics* 2000; **26**: 211-215. **Citations 1150**

- VJ Davies, AJ Hollins, MJ Piechota, W Yip, JR Davies, KE White, PP Nichols, ME Boulton, **M Votruba.** Opa1 deficiency in a mouse model of Autosomal Dominant Optic Atrophy impairs mitochondrial morphology, optic nerve structure and visual function. *Human Molecular Genetics* 2007; **16**: 1307-1318. **Citation 302**
- P Yu-Wai-Man, PG Griffiths, GS Gorman, CM Lourenco, AF Wright, ...Turnbull DM, **Votruba M**, Zeviani M, Carelli V, Bindoff LA, Horvath R, Amati-Bonneau P, Chinnery PF. Multi-system neurological disease is common in patients with OPA1 mutations. *Brain* 2010; **133**: 771-786. **Citations 265**

Publications in last 5 years 2014 - 2018

- Diot A, Agnew T, Sanderson J, Liao C, Carver J, Neves R, Gupta R, Guo Y, Waters C, Seto S, Daniels M, Dombi E, Lodge T, Morten K, Williams S, Enver T, Iborra FJ, **Votruba M**, Poulton J. Validating the RedMIT/GFP-LC3 mouse model by studying mitophagy in Autosomal Dominant Optic Atrophy due to the OPA1Q285STOP mutation. Front Cell Dev Biol. 2018 19;6:103. **Citations 0**
- Jurkute N, Majander A, Bowman R, **Votruba M,** Abbs S, Acheson J, Lenaers G, Amati Bonneau P, Moosajee M, Arno G, Yu Wai Man P. Clinical utility gene card for: inherited optic neuropathies including next-generation sequencing-based approaches. Eur J Hum Genet, 2018. **Citations 0**
- Kousal B., Kolarova H., Meliska M., Bydzovsky J., Diblik P., Kulhanek J., **Votruba M**., Honzik T., and Liskova P. (2018) Peripapillary microcirculation in Leber hereditary optic neuropathy. Acta Ophthalmol. 2018; 26. **Citations 0**
- Mahajan D, **Votruba M**. Can the retina be used to diagnose and plot the progression of Alzheimer's disease? Acta Ophthalmol. 2017;95:768-777. **Citations 6**
- Mahajan D, **Votruba M**. A novel NR2E3 gene mutation in autosomal recessive retinitis pigmentosa with cystic maculopathy. Acta Ophthalmol. 2017; 29. **Citations 1**
- Carelli V, Carbonelli M, de Coo IF, Kawasaki A, Klopstock T, Lagrèze WA, La Morgia C, Newman NJ, Orssaud C, Pott JWR, Sadun AA, van Everdingen J, Vignal-Clermont C, **Votruba M**, Yu-Wai-Man P, Barboni P. International Consensus Statement on the Clinical and Therapeutic Management of Leber Hereditary Optic Neuropathy. J Neuroophthalmol. 2017;37:371-381. **Citations 15**
- Majander A, Robson AG, João C, Holder GE, Chinnery PF, Moore AT, **Votruba M**, Stockman A, Yu-Wai-Man P. The pattern of retinal ganglion cell dysfunction in Leber hereditary optic neuropathy. Mitochondrion. 2017;36:138-149. **Citations 4**
- Beirne K, Rozanowska M, **Votruba M**. Photostimulation of mitochondria as a treatment for retinal neurodegeneration. Mitochondrion. 2017;36:85-95. **Citations 3**
- Majander A, Bowman R, Poulton J, Antcliff RJ, Reddy MA, Michaelides M, Webster AR, Chinnery PF, **Votruba M**, Moore AT, Yu-Wai-Man P. Childhood-onset Leber hereditary optic neuropathy. Br J Ophthalmol. 2017;101:1505-1509. **Citations 6**
- Karaa A, Rahman S, Lombès A, Yu-Wai-Man P, Sheikh MK, Alai-Hansen S, Cohen BH, Dimmock D, Emrick L, Falk MJ, McCormack S, Mirsky D, Moore T, Parikh S, Shoffner J, Taivassalo T, Tarnopolsky M, Tein I, Odenkirchen JC, Goldstein A; **Mito Working Group Member Participants**. Common data elements for clinical research in mitochondrial disease: a National Institute for Neurological Disorders and Stroke project.J Inherit Metab Dis. 2017;40:403-414. **Citations 40**
- Majander A, João C, Rider AT, Henning GB, **Votruba M**, Moore AT, Yu-Wai-Man P, Stockman A. The Pattern of Retinal Ganglion Cell Loss in OPA1-Related Autosomal Dominant Optic Atrophy Inferred From Temporal, Spatial, and Chromatic Sensitivity Losses. Invest Ophthalmol Vis Sci. 2017; 58:502-516. **Citations 3**
- Liao C, Ashley N, Diot A, Morten K, Phadwal K, Williams A, Fearnley I, Rosser L, Lowndes J, Fratter C, Ferguson DJ, Vay L, Quaghebeur G, Moroni I, Bianchi S, Lamperti C, Downes SM, Sitarz KS, Flannery PJ, Carver J, Dombi E, East D, Laura M, Reilly MM, Mortiboys H, Prevo R, Campanella M, Daniels MJ, Zeviani M, Yu-Wai-

Man P, Simon AK, **Votruba M**, Poulton J. Dysregulated mitophagy and mitochondrial organization in optic atrophy due to OPA1 mutations. Neurology. 2017;88(2):131-142. **Citations 21**

- Liskova P, Tesarova M, Dudakova L, Svecova S, Kolarova H, Honzik T, Seto S, **Votruba M**. OPA1 analysis in an international series of probands with bilateral optic atrophy. *Acta Ophthalmol*. 2017; **95**: 363-369. **Citations 4**
- Yu-Wai-Man P, **Votruba M**, Burté F, La Morgia C, Barboni P, Carelli V. A neurodegenerative perspective on mitochondrial optic neuropathies. *Acta Neuropathol*. 2016;132:789-806. **Citations 29**
- Beirne K, Rozanowska MR, **Votruba M**. Red Light Treatment in an Axotomy Model of Neurodegeneration. *Photochemistry & Photobiology*. 2016;**92**:624-31. **Citations 3**
- Kushnareva Y, Seong Y, Andreyev AY, Kuwana T, Kiosses W, **Votruba M**, Newmeyer D D. Mitochondrial dysfunction in an Opa1Q285STOP mouse model of dominant optic atrophy results from Opa1 haploinsufficiency. *Cell Death & Disease,* 2016 **28**;7:e2309. **Citations 8**
- AE Navein, EJ Cooke, JR Davies, A Ohazama, NC Jones, PT Sharpe, SL Evans, BAJ Evans, **M Votruba,** T Wells. Impaired mitochondrial function in the Opa3L122P mouse model for Costeff Syndrome has differential effects on skeletal integrity. *Hum Mol Genet*. 2016; **25**:2404-2416. **Citations 2**
- Smith TG, Seto S, Ganne P, **Votruba M**. A randomized, placebo-controlled trial of the benzoquinone idebenone in a mouse model of OPA1-related dominant optic atrophy reveals a limited therapeutic effect on retinal ganglion cell dendropathy and visual function. *Neuroscience* 2016;**319**:92-106. **Citations 10**
- Morny EK, Margrain TH, Binns AM, **Votruba M**. Electrophysiological ON and OFF Responses in Autosomal Dominant Optic Atrophy. *Invest Ophthalmol Vis Sci*. 2015;**56**:7629-37. **Citations 6**
- Ganne P, **Votruba M.** Retinal changes in a patient with acquired partial lipodystrophy (Laignel-Lavastine and Viard Sndrome). *Acta Ophthalmol*. 2015;**17. Citations 0**
- Ganne P, Garrioch R, **Votruba M.** Perceptions and understanding of genetics and genetic eye disease and attitudes to genetic testing and gene therapy in a primary eye care setting. *Opthalmic Genet.* 2015; **36**:50-7. **Citations 3**
- Sergouniotis PI, Perveen R, Thiselton DL, Giannopoulos K, Sarros M, Davies JR, Biswas S, Ansons AM, Ashworth JL, Lloyd IC, Black GC, **Votruba M.** Clinical and molecular genetic findings in autosomal dominant OPA3 related optic neuropathy. *Neurogenetics*. 2015; **16**:69-75. **Citations 9**
- Smith TG and **Votruba M**. Inherited dominant optic neuropathy: from clinical studies to gene function and back again. *Drug Discovery Today: Disease Models*. 2014;**10**.1016. **Citations 0**
- Rowe F, Wormald R, Cable R, Acton M, Bonstein K, Bowen M, Bronze C, Bunce C, Conroy D, Cowan K, Evans K, Fenton M, Giles H, Gordon I, Halfhide L, Harper R, Lightstone A, **Votruba M**, Waterman H, Zekite A. The Sight Loss and Vision Priority Setting Partnership (SLV-PSP): overview and results of the research prioritisation survey process. *BMJ Open*. 2014; **23**;4:e004905. **Citations 16**
- Yu-Wai-Man P, **Votruba M**, Moore AT, Chinnery PF. Treatment strategies for inherited optic neuropathies: past, present and future. *Eye* (Lond). 2014;**28**:521-37. **Citations 84**

Signed:

Date: 1st February, 2019.