Nicholas **Owen**

Bioinformatician · Research Fellow

nicholas.owen1@gmail.com | nicholas-owen | @Dr__NO | 0000-0001-5598-6274 |

Scientist at the interface of translational research into rare diseases and computational data analysis. Excellent written and verbal communication skills, as exemplified by presentations, scientific posters and high quality peer-reviewed publications. Exceptional time management, handling multiple parallel diverse projects whilst engaging in mentoring, supervision, and academic citizenship activities.

Skills

Data Science/Informatics

- Programming with R for Statistics, Python, Linux and HPC systems for bioinformatic data analysis.
- Expert knowledge of various software packages including bioinformatic tools, data presentation packages, databases, office software and internet tools.
- Creation of modular pipelines including flow and version-control for analysis of RNA-seq, Whole Genome Sequence and epigenetic (RRBS) analysis.
- Generation of Markdown reports for dissemination of results to all levels of multidisclipinary teams.
- Supporter of FAIR principles and Open Science Framework.
- Skilled in all aspects of computer hardware and software, including configuration and maintenance of systems.

Organisation

- Keen eye for detail, applied in accurate record keeping and data management within strict, non-negotiable deadlines.
- Highly skilled in the prioritisation of multi-disciplined workloads for self and others regarding both long term strategy and short-term goals
- Strong ability to train and direct staff and students with a proactive approach within the laboratory.

Communication

- Effective communication skills developed when establishing and maintaining collaborations.
- Skilled in identifying appropriate communication channels to be used in the dissemination of information.
- Proactive approach used effectively when consulting the Head of Laboratory, presenting forward strategies for own group resulting in improved efficiency and morale.
- Skills recognised by scientific community when invited to review manuscripts, as well as through invited presentations at international meetings.
- Written skills developed through the publication of results in high quality, peer reviewed scientific journals in addition to writing scientific and lay reports to grant awarding bodies.

Laboratory

- Excellent ability in a wide range of molecular and cellular biology techniques demonstrated by a good publication record within high impact journals.
- Proven track record in the implementation of novel techniques within the laboratory.
- Committed to the very highest standards of laboratory practice.
- Able to critically and methodically evaluate results/data and interpret accurately.

Experience

Bioinformatician / Research Fellow

University College London

Ocular Omics and Therapeutics - Professor Mariya Moosajee

Apr. 2017 - May 2023

- Analysis of genetic and epigenetic mechanisms underlying rare diseases with a focus on congenital eye defects.
- Experimental design, management and data analysis spanning numerous projects within the group.
- Development of data analysis pipelines using HPC platforms for transcriptomics (RNA-seq), whole genome (WGS) and epigenetics (RRBS).
- Analysis of 100,000 Genomes Project participants' whole genome sequence (WGS) as a member of several Genomics England Clinical Interpretation Partnerships (GeCIPs).
- Bioinformatic pipelines developed with R, Python and Shell, using version control (Github).
- Communication of data to multidisciplinary teams including clinical, lab research and programming groups, through generated reports (RMarkdown) with managed access online data tools and regular meetings.
- Collaboration with research groups within UCL to analyse their projects.
- Advocate for reproducible research through modular pipelines, and supporter of FAIR principles and reproducible research.

Postdoctoral Research Associate

University College London

Splicing Factor related Retinitis Pigmentosa (RP) - Professor Andrew Webster

Apr. 2014 - Apr. 2017

- Investigation into the blood transcriptome (RNA-seq) of rare disease patients with eye disorders.
- Whole genome sequence analysis in collaboration with Genomics England Limited and Cambridge University (BRIDGE/SPEED).
- CRISPR/Cas9 genome editing of retinal related cell lines for the generation of functional model systems.
- Isolation and differentiation of patient iPSC lines to produce retinal pigment epithelium and neural retina.
- Honorary contract with Moorfield's Eye Hospital.

Postdoctoral Research Associate

Queen Mary University of London

Identification of novel interactions of BORIS and Amyloid-β in neurons - Professor Denise Sheer

Oct. 2010 - Nov. 2011

- Novel protein/RNA interactions of BORIS (CTCFL) involved in epigenetic regulation using RIP, coIP, EMSA.
- Characterisation of Amyloid-β multimer complexes, a hallmark of Alzheimer's disease using EMSA, RIP.
- Mammalian Cell Culture; Human Neural Stem Cells and common laboratory lines.
- Protein localisation studies utilising confocal fluorescent microscopy.

Characterisation of the Early (E) Splicesomal Complex

University of Leicester

Characterisation of mammalian spliceosomal E Complex - Dr. Olga Makarova

Oct. 2009 - Oct. 2010

- Whilst developing and submitting a fellowship grant to further enhance the strategies for SMA therapy I carried out research on the isolation and characterisation of the Early (E) spliceosomal complex in mammalian systems using LC-MS/MS proteomics.
- Identified the novel presence of the SMN-associated protein complex.
- Demonstrated E Complex formation is vital for RNA splicing progression.

Senior Postdoctoral Research Associate

University of Leicester

Therapeutic RNA development for Motor Neuron Diseases - Professor Ian Eperon

Sep. 2005 - Sep. 2009

- Optimised RNA bifunctional oligonucleotides (TOES) through use of differing chemistries to increase functional transcript levels; phosphorthiate, 2' O-methyl and LNA modifications.
- Development and optimisation of *in vitro* RNA splicing assays for Spinal Muscular Atrophy.
- Generation of alternative spliced mRNA based *in vivo* dual fluorescent reporter system.
- Characterisation of protein:RNA complexes through immunoprecipitation studies.
- Confocal fluorescent microscopy of oligonucleotide uptake using FRET analysis.
- Functional SELEX of optimal RNA sequences for functional restoration of transcripts.
- Maintenance and transfection of mammalian cell lines; SMA fibroblasts, neurons and common cell types.

Postdoctoral Research Assistant

University of Oxford

Molecular Genetics of Spinal Muscular Atrophy - Professor Kay E. Davies

May 2000 - Aug. 2005

- Generation of novel transgenic murine models expressing mutant isoforms of the SMN protein.
- Isolation and characterisation of primary mouse motor neurons from models.
- Protein Protein Interaction (PPI) studies of the SMN-associated protein complex, specifically the Profilins (PFNs) and novel partners identified through a yeast-2-hybrid screen and microarray analysis.
- Characterisation of SMA Patient mutations in SMN using IP and IF studies.

Research Assistant

University of Oxford

Molecular Genetics of Spinal Muscular Atrophy - Professor Kay E. Davies

Nov. 1994 - Feb. 2000

- D.Phil. on the Molecular Genetics of Spinal Muscular Atrophy 1997 – 2000.
- Deletion analysis of candidate genes in patients and use in prenatal diagnosis.
- Identification and characterisation of the fission yeast *S.pombe* SMN orthologue as a model system for SMA.
- Generation of transgenic mouse models expressing alternative SMN isoforms or patient mutations.
- Physical mapping of the SMA critical genomic region on chromosome 5 using PACs.
- Analysis of SMN mutations on associated proteins using the yeast-2-hybrid system.

Research Assistant

WIMM, University of Oxford

Molecular analysis of FraxE mental retardation - Professor Kay E. Davies

Oct. 1993 - Nov. 1994

- Linkage analysis and population screening of the frequency of $(GCG)_n$ repeats in patients.

Scientific Citizenship

Grants / Funding

- Rare Disease RNA Phenotyping Project Application - NIH Bioresource / UCL.

Leadership / Mentorship

- Co-supervisor of successful PhD and MSc students - UCL / Leicester / Oxford.
- Sponsorship Lead for the UK Eye Genetics Group Annual Meeting 2021 committee - London.
- 2022 UCL FBS Faculty-Wide Poster Session Organizer - UCL.
- Lead for UCL Digital Research Infrastructure EOI application - UCL.
- Mentor for PhD students- UCL / Leicester / Oxford.
- Senior Departmental Teaching Associate (STDA) supporting 30+ D.Phil students - Oxford.

Representation

- Equality Challenge Team - Athena Swan - Institute of Ophthalmology, UCL.
- Patient and Public Involvement and Engagement committee - UCL.
- Postdoctoral researchers representative - Leicester.

Reviewer

- Invited reviewer for various peer-reviewed articles including:
- Bioinformatics, Human Molecular Genetics, Genes

Publications

Manuscripts Submitted for Review

Multi-omics analysis identifies cooperation of transcription factors controlling the specific epithelial cell fate of the corneal epithelium

J. Smits, D. Lima Cunha, J. Qu, N. Owen*, L. Latta, N. Szentmary, B. Seitz, L. Roux, M. Moosajee, D. Aberdam, S. Heeringen, H. Zhou

In Review: EMBO Molecular Medicine (https://doi.org/10.1101/2022.07.13.499857). https://doi.org/10.1101/2022.07.13.499857

Overexpression of extracellular matrix proteins, increased cell death and reduced cell proliferation contribute to the pathophysiology of microphthalmia across in vitro patient-derived models

J. Eintracht, N. Owen*, P. Harding, M. M.

Submitted: EMBO

Variant-specific disruption to SOX2-mediated Notch signalling in *PAX6* patient hiPSC optic vesicles

P. Harding, N. Owen*, J. Eintracht, D. Lima Cunha, M. M.

In Review: Development

Dual molecular effects of constitutional *SF3B2* variants cause a novel dominant spliceosomopathy displaying retinitis pigmentosa or acrofacial dysostosis

C. Van Cauwenbergh, S. Van de Sompele, M. Carron, A. Rey, L. Boutaud, N. Owen*, A. Webster, B. Leroy, E. De Baere

Journal Articles

Loss of the crumbs cell polarity complex disrupts epigenetic transcriptional control and cell cycle progression in the developing retina

N. Owen*, M. Toms, Y. Tian, L. Toualbi, R. Richardson, R. Young, D. Tracey-White, P. Dhami, S. Beck, M. Moosajee

The Journal of Pathology (Jan. 2023). 2023

Identification of Novel Coloboma Candidate Genes through Conserved Gene Expression Analyses across Four Vertebrate Species

N. Owen*+, V. Trejo-Reveles+, B. H. Ching Chan, M. Toms, J. J. Schoenebeck, M. Moosajee, J. Rainger

Biomolecules 13.2 (Feb. 2023) p. 293. 2023

Identification of 4 novel human ocular coloboma genes ANK3, BMPR1B, PDGFRA, and CDH4 through evolutionary conserved vertebrate gene analysis

N. Owen*, M. Toms, R. M. Young, J. Eintracht, H. Sarkar, B. P. Brooks, Genomics-England, M. Moosajee

Genetics in Medicine 0.0 (Jan. 2022) pp. 1–12. 2022

REP1 deficiency causes systemic dysfunction of lipid metabolism and oxidative stress in choroideremia

D. L. Cunha, R. Richardson, D. Tracey-White, A. Abbouda, A. Mitsios, V. Horneffer-van der Sluis, P. Takis, N. Owen*, J. Skinner, A. A. Welch, M. Moosajee

JCI Insight 6.9 (May 2021) p. 146934. 2021

From Transcriptomics to Treatment in Inherited Optic Neuropathies

M. J. Gilhooley, N. Owen*, M. Moosajee, P. Yu Wai Man

Genes 12.2 (Feb. 2021) p. 147. 2021

***EPHA2* Segregates with Microphthalmia and Congenital Cataracts in Two Unrelated Families**

P. Harding, M. Toms, E. Schiff, N. Owen*, S. Bell, I. C. Lloyd, M. Moosajee

International Journal of Molecular Sciences 22.4 (Feb. 2021) p. 2190. 2021

Ocular Phenotype Associated with *DYRK1A* Variants

C. Méjécase, C. M. Way, N. Owen*, M. Moosajee

Genes 12.2 (Feb. 2021) p. 234. 2021

***PAX6* missense variants in two families with isolated foveal hypoplasia and nystagmus: evidence of paternal postzygotic mosaicism**

N. Owen*+, D. Lima Cunha+, V. Tailor, M. Corton, M. Theodorou, M. Moosajee

European Journal of Human Genetics 29.2 (Feb. 2021) pp. 349–355. 2021

Testicular somatic cell-like cells derived from embryonic stem cells induce differentiation of epiblasts into germ cells

H. Rore, N. Owen*, R. E. Piña-Aguilar, K. Docherty, R. Sekido

Communications Biology 4.1 (June 2021) p. 802. 2021

Zebrafish retinal mRNA RNA-seq data processing

N. Owen*, M. Moosajee

ProtocolExchange (May 2020). 2020

Data Sharing: a Primer from UKRN

J. N. Towse, S. Rumsey, N. Owen*, P. Langford, M. Jaquiery, C. Bolibaugh

(Oct. 2020). OSF Preprints, 2020

Open Code and Software: a Primer from UKRN

A. Turner, M. Topor, N. Owen*, A. J. Stewart, A. R. Kenny, A. L. Jones, D. A. Ellis

(Oct. 2020). OSF Preprints, 2020

RNA-sequencing in ophthalmology research: considerations for experimental design and analysis

N. Owen*, M. Moosajee

Therapeutic Advances in Ophthalmology 11 (Jan. 2019) p. 2515841419835460. 2019

Transcriptome profiling of zebrafish optic fissure fusion

R. Richardson, N. Owen*, M. Toms, R. M. Young, D. Tracey-White, M. Moosajee

Scientific Reports 9.1 (Feb. 2019) p. 1541. 2019

Genome-wide RNA-Sequencing analysis identifies a distinct fibrosis gene signature in the conjunctiva after glaucoma surgery

C. Yu-Wai-Man, N. Owen*, J. Lees, A. D. Tagalakis, S. L. Hart, A. R. Webster, C. A. Orengo, P. T. Khaw

Scientific Reports 7.1 (July 2017) p. 5644. 2017

Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene *IFT140*

S. Hull, N. Owen*, F. Islam, D. Tracey-White, V. Plagnol, G. E. Holder, M. Michaelides, K. Carss, F. L. Raymond, J.-M. Rozet, S. C. Ramsden, G. C. M. Black, I. Perrault, A. Sarkar, M. Moosajee, A. R. Webster, G. Arno, A. T. Moore

Investigative Ophthalmology & Visual Science 57.3 (Mar. 2016) pp. 1053–1062. 2016

BORIS/CTCFL is an RNA-binding protein that associates with polysomes

B. W. Ogunkolade, T. A. Jones, J. Aarum, J. Szary, N. Owen*, D. Ottaviani, M. A. Mumin, S. Patel, C. A. Pieri, A. R. Silver, D. Sheer

BMC Cell Biology 14.1 (Nov. 2013) p. 52. 2013

Functional mammalian spliceosomal complex E contains SMN complex proteins in addition to U1 and U2 snRNPs

E. M. Makarov, N. Owen*, A. Bottrill, O. V. Makarova

Nucleic Acids Research 40.6 (Mar. 2012) pp. 2639–2652. 2012

Design principles for bifunctional targeted oligonucleotide enhancers of splicing

N. Owen*, H. Zhou, A. A. Malygin, J. Sangha, L. D. Smith, F. Muntoni, I. C. Eperon

Nucleic Acids Research 39.16 (Sept. 2011) pp. 7194–7208. 2011

Characterization of the Schizosaccharomyces pombe orthologue of the human survival motor neuron (SMN) protein

N. Owen*, C. L. Doe, J. Mellor, K. E. Davies

Human Molecular Genetics 9.5 (Mar. 2000) pp. 675–684. 2000

Analysis of mutations in the tudor domain of the survival motor neuron protein SMN

P. Mohaghegh, N. R. Rodrigues, N. Owen*, C. P. Ponting, T. T. Le, A. H. Burghes, K. E. Davies

European journal of human genetics: EJHG 7.5 (July 1999) pp. 519–525. 1999

Gene deletions in spinal muscular atrophy.

N. R. Rodrigues, N. Owen*, K. Talbot, S. Patel, F. Muntoni, J. Ignatius, V. Dubowitz, K. E. Davies

Journal of Medical Genetics 33.2 (Feb. 1996) pp. 93–96. 1996

A member of the MAP kinase phosphatase gene family in mouse containing a complex trinucleotide repeat in the coding region

A. M. Theodosiou, N. R. Rodrigues, M. A. Nesbit, H. J. Ambrose, H. Paterson, E. McLellan-Arnold, Y. Boyd, M. A. Leversha, N. Owen*, D. J. Blake, A. Ashworth, K. E. Davies

Human Molecular Genetics 5.5 (May 1996) pp. 675–684. 1996

Prenatal diagnosis of spinal muscular atrophy by gene deletion analysis

N. R. Rodrigues, L. Campbell, N. Owen*, C. H. Rodeck, K. E. Davies

Lancet 345.8956 (Apr. 1995) p. 1049. 1995

Deletions in the survival motor neuron gene on 5q13 in autosomal recessive spinal muscular atrophy

N. Rodrigues, N. Owen*, K. Talbot, J. Ignatius, V. Dubowitz, K. Davies

Human Molecular Genetics 4.4 (Apr. 1995) pp. 631–634. 1995

Presentations - Recent

Identification of novel human ocular coloboma genes through evolutionary conserved vertebrate gene analysis — *London, U.K.*
Genomics England Research Meeting — *June 2022*

Pros and cons of single-cell versus bulk RNA-seq — *London, U.K.*
Single Cell Omics Workshop 2022 — *June 2022*

***BMPR1B* identified as a novel human ocular coloboma gene through cross-species meta-analysis** — *Denver, U.S.A.*
ARVO 2022 Annual Meeting — *May 2022*

Omics of Eye Disease: from models to patients — *London, U.K.*
Institute of Ophthalmology Research Network Seminar — *Feb. 2021*

Unravelling the process of optic fissure closure through spatial and temporal transcriptome analysis in zebrafish — *London, U.K.*
IOO Research Day — *Jul. 2020*

Education

D.Phil in Molecular Genetics of Spinal Muscular Atrophy - Prof. Kay E. Davies — *Oxford, UK*
University of Oxford, Department of Human Anatomy and Genetics — *2000*

M.Sc. Human Molecular Genetics — *London, UK*
St. Marys Hospital Medical School — *1993*

B.Sc. Hons. Genetics — *London, UK*
Queen Mary University of London — *1992*

Memberships

2021-	**International Society for Computational Biology**, Member	*Virginia, U.S.A.*
2020-	**Association for Research in Vision and Ophthalmology (ARVO)**, Member	*Maryland, U.S.A*
1996-	**American Society of Human Genetics (ASHG)**, Member	*Las Vegas, U.S.A*
1996-	**The RNA Society**, Member	*Maryland, U.S.A.*
1998-	**The Genetics Society**, Member	*U.K.*
2020-	**The U.K. Eye Genetics Group**, Member	*U.K.*
2016-	**Genomics England Clinical Interpretation Partnership (GeCIPs)**, Member **Hearing/Sight, Neuroscience, Transcriptomics/Machine Learning**,	*U.K.*
2018-	**NHS-R Community**, Member	*U.K.*