

## Curriculum Vitae

**Name:** Dr. Anna M. Rose  
**Date of Birth:** 12/01/1987  
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**EDUCATION:** 1997-2005 Dame Alice Owen's School  
2005-present MBPhD student, University College London Medical School

### ACADEMIC QUALIFICATIONS

**PhD in Human Molecular Genetics – October 2012**  
**University College London**

Thesis title – “Transcriptional regulation of *PRPF31*: the role of variable expression in determining phenotype in autosomal dominant retinitis pigmentosa”

**BSc (Hons) – June 2009**  
**University College London**

Medical Sciences with Anatomy and Developmental Biology  
1<sup>st</sup> class (starred)

### UNIVERSITY ACADEMIC AWARDS

UCL Year 1 Medical Sciences Merit Award – May 2006

UCL Pre-clinical Medical Sciences Distinction Award – May 2007

Division of Biosciences Deans List for Academic Excellence 2009

Division of Biosciences Faculty Medal 2008-9 Nominee

Cordwainer's Prize for Best MBPhD thesis – January 2013

Allen Goldsmith Prize for Cancer Medicine – June 2013

John Jepson Memorial Prize for the Scientific Basis of Medicine – June 2013

### NATIONAL ACADEMIC AWARDS

Royal Society of Medicine (Ophthalmology Section and Student Members Group) UCO 2009 Best Case Presentation – April 2009

Royal College of Ophthalmologists' Duke Elder Prize Examination – May 2009

Royal Society of Medicine Student Research Prize (First Place) – November 2009

Royal Society of Medicine Brigadier Haywood prize (shortlisted) – February 2010

Royal College of Paediatrics and Child Health Tony Jackson Prize – January 2012

Medical Acorn Foundation Conference Best Oral Presentation (Medicine) – October 2012

Rosetrees Trust 25<sup>th</sup> Anniversary Symposium Best Poster Presentation – October 2012

Royal College of Ophthalmologists' Duke Elder Prize Examination – May 2013

### **ABSTRACT PUBLICATIONS**

**Rose AM**, Webster AR, Mukhopadhyay R, Bhattacharya SS, Waseem NH. Identification of Large Deletions in *PRPF31* in Autosomal Dominant Retinitis Pigmentosa. IOVS 2010; 51:ARVO E-Abstract 4093

### **ARTICLE PUBLICATIONS**

**Rose AM**, Gradin DS, Mundia D. *Bilateral lens subluxation in a patient with suspected Loeys-Dietz syndrome*. Afr J Paediatr Surg 2011; 8:119-20.

**Rose AM**, Kabiru J, Rose GE. *Alveolar soft-part sarcoma of the orbit*. Afr J Paediatr Surg 2011; 8:82-4.

**Rose AM**, Mukhopadhyay R, Webster AR, Bhattacharya SS, Waseem NH. *A 112kb deletion in chromosome 19q13.42 leads to retinitis pigmentosa*. IOVS 2011; 52:6597-6603.

Mitchison NA, **Rose AM**. *Epistasis: The key to understanding immunological disease?* Eur J Immunol 2011; 41:2152–2154.

**Rose AM**, Shah AZ, Waseem NH, Chakarova CF, Alfano G, Coussa RG, Ajlan R, Koenekoop RK, Bhattacharya SS. *Expression of PRPF31 and TFPT: regulation in health and retinal disease*. Hum Mol Genet 2012; 21:4126-37.

**Rose AM**, Bell LCK. *Epistasis and immunity: the role of genetic interactions in autoimmune diseases*. Immunology 2012; 137:131-8.

Venturini G, **Rose AM**, Shah AZ, Bhattacharya SS, Rivolta C. *CNOT3 is a modifier of PRPF31 mutations in retinitis pigmentosa with incomplete penetrance*. PLoS Genetics 2012; 8(11):e1003040

**Rose AM**, Kabiru J, Rose GE. *A rare case of orbital haemangiopericytoma arising in childhood*. Orbit 2013; 32(6):384-6.

**Rose AM**, Shah AZ, Alfano G, Bujakowska KM, Barker AF, Robertson JL, Rahman S, Valdez Sanchez L, Diaz-Corrales F, Chakarova CF, Krishna A, Bhattacharya SS. *A study into the evolutionary divergence of the core promoter elements of PRPF31 and TFPT*. J Mol Genet Med 2013; 7:67.

**Rose AM**, Shah AZ, Venturini G, Rivolta C, Rose GE, Bhattacharya SS. *Dominant PRPF31 mutations are hypostatic to a recessive CNOT3 polymorphism in retinitis pigmentosa: A Novel Phenomenon of "Linked Trans-Acting Epistasis."* Annals of Human Genetics 2013; in press.

**Rose AM**, Hall CS, Martinez-Alier N. *Aetiology and management of malnutrition in HIV-positive children*. Archives of Disease in Childhood 2014; ePub ahead of print.

## **MANUSCRIPTS IN PREPARATION**

**Rose AM**, Barnett J, Morris-Jones S, Marks DJB. Staphylococcus lugdunensis septic arthritis and epidural abscess in a patient receiving anti-TNF therapy. *Rheumatology – under review*.

**Rose AM**, Alfano G, Muspratt-Tucker N, Moore AT, Bhattacharya SS, Webster AR. *Diverse clinical phenotypes associated with Arg437X mutation in FAM161A*.

## **PEER REVIEW EXPERIENCE**

I have acted as a peer-reviewer for the following journals and funding bodies:

- International Journal of Neuroscience
- Journal of Rheumatology
- BMC Research Notes
- Indian Journal of Ophthalmology
- Rosetrees Trust
- Medical Research Council (South Africa)

## **RESEARCH INTERESTS**

My primary area of research concerns autosomal dominant retinitis pigmentosa, principally the form associated with mutations in the *PRPF31* gene. My current work involves understanding the mechanism underlying control of gene expression and its' relation to clinical presentation of disease. More broadly, I am interested in the role that genes play in human health and disease and I have published work in several areas, including clinical presentation of congenital genetic syndromes, oncogenetics and immunogenetics.

## **MEDICAL WORK EXPERIENCE**

**July-September 2006:** 3 months auxiliary nursing in the Accident and Emergency Department of Moorfield's Eye Hospital. This work included taking patient's medical histories, simple clinical procedures such as measurement of blood glucose, blood pressure and a whole range of vision tests; for instance, visual acuity and Ishihara colour vision charts. I also assisted in the Minor Operations Theatre, cared for patients post-operatively in the Observation Bays and generally tried to maintain the well-being of the patients.

**July - August 2007:** Two months voluntary work in Livingstone, Zambia with Rainbow Trust Africa, working with the Mwenda Medical Centre team. This involved examining and assessing patients in both an urban and rural medical setting, assisting with childhood vaccination programmes and working in the contraceptive advice clinic. I also undertook research for the charity into HIV/AIDS awareness amongst 13-14 year old school children, with a view to improving sex education in the charity-run school.

**June-July 2009:** 7 weeks work with Rainbow Trust Africa in Livingstone, Zambia. The main focus of this work was conducting research into morbidity and mortality amongst HIV positive children, with over 500 hospital records reviewed. This involved working closely with the Ministry of Health and Livingstone Government Hospital. The aim of the research was to establish whether a need existed for a paediatric respite and hospice facility at the charity's main

site in Livingstone. I also spent time with other healthcare providers, such as St. Joseph's Hospice and the SEPO HIV/AIDS centre.

### **OTHER INTERESTS**

I am a keen musician and am involved in the RUMS Music Society, as well as the London Gallery Quire. I enjoy performing as a soloist, as well as in chamber groups. I also row for the senior squad of my medical school boat club and take great pleasure in competing in this sport.

### **REFERENCES**

**Prof. S. Bhattacharya**

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**Prof. G. Stewart**

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